Gabriele Siciliano

List of Publications by Year
in descending order

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394 papers	13,750 citations	26630 56 h-index	36028 97 g-index
404 all docs	404 docs citations	404 times ranked	18482 citing authors

#	Article	IF	Citations
1	Common variants at ABCA7, MS4A6A/MS4A4E, EPHA1, CD33 and CD2AP are associated with Alzheimer's disease. Nature Genetics, 2011, 43, 429-435.	21.4	1,708
2	APOE and Alzheimer disease: a major gene with semi-dominant inheritance. Molecular Psychiatry, 2011, 16, 903-907.	7.9	529
3	Cytochrome c oxidase and mitochondrial F1F0-ATPase (ATP synthase) activities in platelets and brain from patients with Alzheimer's disease. Neurobiology of Aging, 2002, 23, 371-376.	3.1	333
4	Causative and susceptibility genes for Alzheimerâ€™s disease: a review. Brain Research Bulletin, 2003, 61, 1-24.	3.0	267
5	Oxidative DNA damage in peripheral leukocytes of mild cognitive impairment and AD patients. Neurobiology of Aging, 2005, 26, 567-573.	3.1	252
6	Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. Nature Genetics, 2021, 53, 1636-1648.	21.4	223
7	Executive dysfunction and avoidant personality trait in myotonic dystrophy type 1 (DM-1) and in proximal myotonic myopathy (PROMM/DM-2). Neuromuscular Disorders, 2003, 13, 813-821.	0.6	198
8	EFNS guidelines on the diagnostic approach to pauciâ€•or asymptomatic hyperCKemia. European Journal of Neurology, 2010, 17, 767-773.	3.3	157
9	Phenotypic heterogeneity of the 8344A>G mtDNA â€œMERRFâ€•mutation. Neurology, 2013, 80, 2049-2054.	1.1	157
10	Mutations of FUS gene in sporadic amyotrophic lateral sclerosis. Journal of Medical Genetics, 2010, 47, 190-194.	3.2	152
11	Multiâ€•center assessment of the Total Neuropathy Score for chemotherapyâ€•induced peripheral neurotoxicity. Journal of the Peripheral Nervous System, 2006, 11, 135-141.	3.1	141
12	A genome-wide association meta-analysis identifies a novel locus at 17q11.2 associated with sporadic amyotrophic lateral sclerosis. Human Molecular Genetics, 2014, 23, 2220-2231.	2.9	123
13	The m.3243A>G mitochondrial DNA mutation and related phenotypes. A matter of gender?. Journal of Neurology, 2014, 261, 504-510.	3.6	119
14	Searching for the role and the most suitable biomarkers of oxidative stress in Alzheimer's disease and in other neurodegenerative diseases. Neurobiology of Aging, 2005, 26, 587-595.	3.1	118
15	Observational clinical study in juvenile-adult glycogenosis type 2 patients undergoing enzyme replacement therapy for up to 4Âyears. Journal of Neurology, 2012, 259, 952-958.	3.6	117
16	A standardized clinical evaluation of patients affected by facioscapulohumeral muscular dystrophy: The FSHD clinical score. Muscle and Nerve, 2010, 42, 213-217.	2.2	113
17	Phenotypic manifestations associated with CAG-repeat expansion in the androgen receptor gene in male patients and heterozygous females: a clinical and molecular study of 30 families. Neuromuscular Disorders, 2000, 10, 391-397.	0.6	112
18	Molecular diagnosis in LGMD2A: Mutation analysis or protein testing?. Human Mutation, 2004, 24, 52-62.	2.5	109

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19	Atypical CIDP: diagnostic criteria, progression and treatment response. Data from the Italian CIDP Database. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 125-132.	1.9	108
20	A two-stage genome-wide association study of sporadic amyotrophic lateral sclerosis. Human Molecular Genetics, 2009, 18, 1524-1532.	2.9	106
21	Large-Scale Population Analysis Challenges the Current Criteria for the Molecular Diagnosis of Fascioscapulohumeral Muscular Dystrophy. American Journal of Human Genetics, 2012, 90, 628-635.	6.2	104
22	Genome-wide haplotype association study identifies the FRMD4A gene as a risk locus for Alzheimer's disease. Molecular Psychiatry, 2013, 18, 461-470.	7.9	103
23	Phenotype modulators in myophosphorylase deficiency. Annals of Neurology, 2003, 53, 497-502.	5.3	101
24	Minimally invasive approach for adrenal lesions: Systematic review of laparoscopic versus retroperitoneoscopic adrenalectomy and assessment of risk factors for complications. International Journal of Surgery, 2016, 28, S118-S123.	2.7	99
25	Nusinersen safety and effects on motor function in adult spinal muscular atrophy type 2 and 3. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 1166-1174.	1.9	99
26	Brainstem neurodegeneration correlates with clinical dysfunction in SCA1 but not in SCA2. A quantitative volumetric, diffusion and proton spectroscopy MR study. Brain, 2004, 127, 1785-1795.	7.6	98
27	Autosomal dominant external ophthalmoplegia and bipolar affective disorder associated with a mutation in the ANT1 gene. Neuromuscular Disorders, 2003, 13, 162-165.	0.6	95
28	Central nervous system involvement in Anderson-Fabry disease: a clinical and MRI retrospective study. Journal of Neurology, Neurosurgery and Psychiatry, 2008, 79, 1249-1254.	1.9	95
29	TARDBP (TDPâ€43) sequence analysis in patients with familial and sporadic ALS: identification of two novel mutations. European Journal of Neurology, 2009, 16, 727-732.	3.3	93
30	SOD1 mutations in amyotrophic lateral sclerosis. Journal of Neurology, 2005, 252, 782-788.	3.6	86
31	The italian limb girdle muscular dystrophy registry: Relative frequency, clinical features, and differential diagnosis. Muscle and Nerve, 2017, 55, 55-68.	2.2	86
32	Large scale genotypeâ€“phenotype analyses indicate that novel prognostic tools are required for families with facioscapulohumeral muscular dystrophy. Brain, 2013, 136, 3408-3417.	7.6	85
33	Pleiotropic effects of spastin on neurite growth depending on expression levels. Journal of Neurochemistry, 2009, 108, 1277-1288.	3.9	84
34	Coenzyme Q10 is frequently reduced in muscle of patients with mitochondrial myopathy. Neuromuscular Disorders, 2010, 20, 44-48.	0.6	84
35	Evidence of the association of BIN1 and PICALM with the AD risk in contrasting European populations. Neurobiology of Aging, 2011, 32, 756.e11-756.e15.	3.1	82
36	Clinical and Molecular Characterization of Patients With Limb-Girdle Muscular Dystrophy Type 2I. Archives of Neurology, 2005, 62, 1894.	4.5	81

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37	Evaluating the levels of interleukin-1 family cytokines in sporadic amyotrophic lateral sclerosis. Journal of Neuroinflammation, 2014, 11, 94.	7.2	74
38	From mild cognitive impairment to dementia: a prevalence study in a district of Tuscany, Italy. Acta Neurologica Scandinavica, 2005, 112, 65-71.	2.1	72
39	Decreased platelet cytochrome c oxidase activity is accompanied by increased blood lactate concentration during exercise in patients with Alzheimer disease. Experimental Neurology, 2003, 182, 421-426.	4.1	70
40	Structural and functional evaluation of cortical motor areas in Amyotrophic Lateral Sclerosis. Experimental Neurology, 2012, 234, 169-180.	4.1	70
41	Redefining phenotypes associated with mitochondrial DNA single deletion. Journal of Neurology, 2015, 262, 1301-1309.	3.6	68
42	Magnetic susceptibility in the deep layers of the primary motor cortex in Amyotrophic Lateral Sclerosis. NeuroImage: Clinical, 2016, 12, 965-969.	2.7	68
43	Could mitochondrial haplogroups play a role in sporadic amyotrophic lateral sclerosis?. Neuroscience Letters, 2004, 371, 158-162.	2.1	67
44	Oxidative stress biomarkers in patients with untreated obstructive sleep apnea syndrome. Sleep Medicine, 2012, 13, 632-636.	1.6	67
45	Decreased Methylation of the Mitochondrial D-Loop Region in Late-Onset Alzheimerâ€™s Disease. Journal of Alzheimer's Disease, 2017, 59, 559-564.	2.6	66
46	Oxidative stress biomarkers in mitochondrial myopathies, basally and after cysteine donor supplementation. Journal of Neurology, 2010, 257, 774-781.	3.6	65
47	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
48	Association of maternal polymorphisms in folate metabolizing genes with chromosome damage and risk of Down syndrome offspring. Neuroscience Letters, 2009, 449, 15-19.	2.1	63
49	Genetic characterization in symptomatic female DMD carriers: lack of relationship between X-inactivation, transcriptional DMD allele balancing and phenotype. BMC Medical Genetics, 2012, 13, 73.	2.1	63
50	Methylation analysis of multiple genes in blood DNA of Alzheimerâ€™s disease and healthy individuals. Neuroscience Letters, 2015, 600, 143-147.	2.1	63
51	Disease awareness in myotonic dystrophy type 1: an observational cross-sectional study. Orphanet Journal of Rare Diseases, 2016, 11, 34.	2.7	63
52	Folate, Homocysteine, Vitamin B12, and Polymorphisms of Genes Participating in One-Carbon Metabolism in Late-Onset Alzheimer's Disease Patients and Healthy Controls. Antioxidants and Redox Signaling, 2012, 17, 195-204.	5.4	60
53	Clinical expression of facioscapulohumeral muscular dystrophy in carriers of 1â€“3 D4Z4 reduced alleles: experience of the FSHD Italian National Registry. BMJ Open, 2016, 6, e007798.	1.9	60
54	Low prevalence of TT virus in the cerebrospinal fluid of viremic patients with central nervous system disorders. Journal of Medical Virology, 2001, 65, 418-422.	5.0	59

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55	Subcutaneous immunoglobulin in CIDP and MMN: a short-term nationwide study. Journal of Neurology, 2014, 261, 2159-2164.	3.6	59
56	Amyotrophic Lateral Sclerosis and Oxidative Stress: A Double-Blind Therapeutic Trial After Curcumin Supplementation. CNS and Neurological Disorders - Drug Targets, 2018, 17, 767-779.	1.4	59
57	Long-term treatment with idebenone and riboflavin in a patient with MELAS. Neurological Sciences, 2000, 21, S981-S981.	1.9	58
58	Risk of arrhythmia in type I myotonic dystrophy: the role of clinical and genetic variables. Journal of Neurology, Neurosurgery and Psychiatry, 2009, 80, 790-793.	1.9	58
59	Differential effects of alendronate and losartan therapy on osteopenia and aortic aneurysm in mice with severe Marfan syndrome. Human Molecular Genetics, 2010, 19, 4790-4798.	2.9	58
60	Detection and quasispecies analysis of hepatitis C virus in the cerebrospinal fluid of infected patients. Journal of NeuroVirology, 1999, 5, 319-323.	2.1	57
61	Muscle modifications in Parkinson's disease: myoelectric manifestations. Electroencephalography and Clinical Neurophysiology - Electromyography and Motor Control, 1996, 101, 211-218.	1.4	56
62	New motor outcome function measures in evaluation of Lateâ€Onset Pompe disease before and after enzyme replacement therapy. Muscle and Nerve, 2012, 45, 831-834.	2.2	56
63	Prognostic factors in mild dystrophinopathies. Journal of the Neurological Sciences, 1996, 142, 70-78.	0.6	55
64	Human dental pulp stem cells protect mouse dopaminergic neurons against MPP+ or rotenone. Brain Research, 2011, 1367, 94-102.	2.2	55
65	A novel clinical tool to classify facioscapulohumeral muscular dystrophy phenotypes. Journal of Neurology, 2016, 263, 1204-1214.	3.6	55
66	Charcot-Marie-Tooth disease type I and related demyelinating neuropathies: Mutation analysis in a large cohort of Italian families. Human Mutation, 2001, 18, 32-41.	2.5	54
67	Mitochondria, Mitochondrial DNA and Alzheimers Disease. What Comes First?. Current Alzheimer Research, 2008, 5, 457-468.	1.4	54
68	The CALHM1 P86L Polymorphism is a Genetic Modifier of Age at Onset in Alzheimer's Disease: a Meta-Analysis Study. Journal of Alzheimer's Disease, 2010, 22, 247-255.	2.6	54
69	Genotype-phenotype correlation in Pompe disease, a step forward. Orphanet Journal of Rare Diseases, 2014, 9, 102.	2.7	54
70	Antioxidant capacity and protein oxidation in cerebrospinal fluid of amyotrophic lateral sclerosis. Journal of Neurology, 2007, 254, 575-580.	3.6	53
71	A Mutation in theCASQ1Gene Causes a Vacuolar Myopathy with Accumulation of Sarcoplasmic Reticulum Protein Aggregates. Human Mutation, 2014, 35, 1163-1170.	2.5	53
72	Antimyoclonic effect of levetiracetam in MERRF syndrome. Journal of the Neurological Sciences, 2006, 243, 97-99.	0.6	52

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73	Relationship between neuropsychological impairment and grey and white matter changes in adult-onset myotonic dystrophy type 1. NeuroImage: Clinical, 2016, 12, 190-197.	2.7	51
74	Identification and characterization of three novel mutations in theCASQ1gene in four patients with tubular aggregate myopathy. Human Mutation, 2017, 38, 1761-1773.	2.5	51
75	Î±-Synuclein Heterocomplexes with Î²-Amyloid Are Increased in Red Blood Cells of Parkinsonâ€™s Disease Patients and Correlate with Disease Severity. Frontiers in Molecular Neuroscience, 2018, 11, 53.	2.9	51
76	LOPED study: looking for an early diagnosis in a late-onset Pompe disease high-risk population. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, jnnp-2014-310164.	1.9	50
77	CSF phosporylated TAU protein levels correlate with cerebral glucose metabolism assessed with PET in Alzheimer's disease. Brain Research Bulletin, 2008, 76, 80-84.	3.0	49
78	POLG1-Related and other â€œMitochondrial Parkinsonismsâ€• an Overview. Journal of Molecular Neuroscience, 2011, 44, 17-24.	2.3	49
79	Epidemiology of myotonic dystrophy in Italy: re-apprisal after genetic diagnosis. Clinical Genetics, 2002, 59, 344-349.	2.0	48
80	Mitochondrial Cascade Hypothesis of Alzheimer's Disease: Myth or Reality?. Antioxidants and Redox Signaling, 2007, 9, 1631-1646.	5.4	48
81	Italian validation of INQoL, a quality of life questionnaire for adults with muscle diseases. European Journal of Neurology, 2010, 17, 1178-1187.	3.3	48
82	Clinical Trials for Neuroprotection in ALS. CNS and Neurological Disorders - Drug Targets, 2010, 9, 305-313.	1.4	48
83	Serotoninergic Polymorphisms (5-HTTLPRand5-HT2A): Association Studies with Psychosis in Alzheimer Disease. Genetic Testing and Molecular Biomarkers, 2003, 7, 309-314.	1.7	47
84	Psychiatric involvement in adult patients with mitochondrial disease. Neurological Sciences, 2013, 34, 71-74.	1.9	47
85	Nusinersen treatment and cerebrospinal fluid neurofilaments: An explorative study on Spinal Muscular Atrophy type 3 patients. Journal of Cellular and Molecular Medicine, 2020, 24, 3034-3039.	3.6	47
86	High-Resolution 7T MR Imaging of the Motor Cortex in Amyotrophic Lateral Sclerosis. American Journal of Neuroradiology, 2016, 37, 455-461.	2.4	46
87	Molecular characterization of myophosphorylase deficiency in a group of patients from Northern Italy. Journal of the Neurological Sciences, 1996, 137, 14-19.	0.6	45
88	A Ser326Cys polymorphism in the DNA repair gene hOGG1 is not associated with sporadic Alzheimer's disease. Neuroscience Letters, 2007, 414, 282-285.	2.1	45
89	Facioscapulohumeral muscular dystrophy: hearing loss and other atypical features of patients with large 4q35 deletions. European Journal of Neurology, 2008, 15, 1353-1358.	3.3	45
90	Effects of aerobic training on lactate and catecholaminergic exercise responses in mitochondrial myopathies. Neuromuscular Disorders, 2000, 10, 40-45.	0.6	43

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91	Functional Diagnostics in Mitochondrial Diseases. Bioscience Reports, 2007, 27, 53-67.	2.4	43
92	The path to biomarker-based diagnostic criteria for the spectrum of neurodegenerative diseases. Expert Review of Molecular Diagnostics, 2020, 20, 421-441.	3.1	42
93	Lack of association between mtDNA haplogroups and Alzheimerâ€™s disease in Tuscany. Neurological Sciences, 2007, 28, 142-147.	1.9	41
94	Absence of angiogenic genes modification in Italian ALS patients. Neurobiology of Aging, 2008, 29, 314-316.	3.1	41
95	Allelic and phenotypic heterogeneity in 49 Italian patients with the muscle form of CPTâ€II deficiency. Clinical Genetics, 2012, 82, 232-239.	2.0	40
96	Oxidative stress and APO E polymorphisms in Alzheimer's disease and in mild cognitive impairment. Free Radical Research, 2013, 47, 569-576.	3.3	40
97	Oxidative Stress Treatment for Clinical Trials in Neurodegenerative Diseases. Journal of Alzheimer's Disease, 2011, 24, 111-126.	2.6	39
98	Exploring the clinical association between neurological symptoms and COVID-19 pandemic outbreak: a systematic review of current literature. Journal of Neurology, 2021, 268, 1561-1569.	3.6	39
99	Thalidomide for improving cutaneous and pulmonary sarcoidosis in patients resistant or with contraindications to corticosteroids. Biomedicine and Pharmacotherapy, 2012, 66, 300-307.	5.6	38
100	Fatigue and exercise intolerance in mitochondrial diseases. Literature revision and experience of the Italian Network of mitochondrial diseases. Neuromuscular Disorders, 2012, 22, S226-S229.	0.6	38
101	â€œMitochondrial neuropathiesâ€: A survey from the large cohort of the Italian Network. Neuromuscular Disorders, 2016, 26, 272-276.	0.6	37
102	Subcutaneous immunoglobulin in CIDP and MMN: a different long-term clinical response?. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 791-793.	1.9	37
103	Association of the hOGG1 Ser326Cys polymorphism with sporadic amyotrophic lateral sclerosis. Neuroscience Letters, 2007, 420, 163-168.	2.1	36
104	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.	0.7	36
105	Impaired oxidative metabolism in exercising muscle from ALS patients. Journal of the Neurological Sciences, 2001, 191, 61-65.	0.6	35
106	Mitochondrial DNA rearrangements in young onset parkinsonism: two case reports. Journal of Neurology, Neurosurgery and Psychiatry, 2001, 71, 685-687.	1.9	35
107	Polymorphisms in folate and homocysteine metabolizing genes and chromosome damage in mothers of Down syndrome children. American Journal of Medical Genetics, Part A, 2007, 143A, 2006-2015.	1.2	35
108	Screening of the PFN1 gene in sporadic amyotrophic lateral sclerosis and in frontotemporal dementia. Neurobiology of Aging, 2013, 34, 1517.e9-1517.e10.	3.1	35

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109	Robotic gait training improves motor skills and quality of life in hereditary spastic paraplegia. NeuroRehabilitation, 2015, 36, 93-99.	1.3	35
110	Impaired oxidative metabolism and lipid peroxidation in exercising muscle from ALS patients. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders: Official Publication of the World Federation of Neurology, Research Group on Motor Neuron Diseases, 2002, 3, 57-62.	1.2	34
111	Activity of protein phosphatase calcineurin is decreased in sporadic and familial amyotrophic lateral sclerosispatients. Journal of Neurochemistry, 2004, 90, 1237-1242.	3.9	34
112	Twinkle mutation in an Italian family with external progressive ophthalmoplegia and parkinsonism: A case report and an update on the state of art. Neuroscience Letters, 2013, 556, 1-4.	2.1	34
113	Plasmatic oxidative stress biomarkers in multiple sclerosis: Relation with clinical and demographic characteristics. Clinical Biochemistry, 2015, 48, 19-23.	1.9	34
114	Coenzyme Q10, exercise lactate and CTG trinucleotide expansion in myotonic dystrophy. Brain Research Bulletin, 2001, 56, 405-410.	3.0	33
115	Myoclonus in mitochondrial disorders. Movement Disorders, 2014, 29, 722-728.	3.9	33
116	Different Clinical Contexts of Use of Blood Neurofilament Light Chain Protein in the Spectrum of Neurodegenerative Diseases. Molecular Neurobiology, 2020, 57, 4667-4691.	4.0	33
117	The unfolded protein response in amyotrophic later sclerosis: results of a phase 2 trial. Brain, 2021, 144, 2635-2647.	7.6	33
118	Novel sarcoglycan gene mutations in a large cohort of Italian patients. Journal of Medical Genetics, 2003, 40, 67e-67.	3.2	32
119	Coenzyme Q10 and Neurological Diseases. Pharmaceuticals, 2009, 2, 134-149.	3.8	32
120	Low frequency stimulation of the nucleus tegmenti pedunculopontini increases cortical metabolism in Parkinsonian patients. European Journal of Neurology, 2011, 18, 842-849.	3.3	32
121	Erythropoietin in amyotrophic lateral sclerosis: a multicentre, randomised, double blind, placebo controlled, phase III study. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 879-886.	1.9	32
122	Î±-Synuclein Aggregates with Î²-Amyloid or Tau in Human Red Blood Cells: Correlation with Antioxidant Capability and Physical Exercise in Human Healthy Subjects. Molecular Neurobiology, 2018, 55, 2653-2675.	4.0	32
123	Autoimmune limbic encephalitis related to SARS-CoV-2 infection: Case-report and review of the literature. Brain, Behavior, & Immunity - Health, 2021, 12, 100210.	2.5	32
124	Progressive sensorineural hearing loss in childhood. Pediatric Neurology, 1999, 20, 130-136.	2.1	31
125	Lactate production and catecholamine profile during aerobic exercise in normotensive OSAS patients. Sleep Medicine, 2004, 5, 137-145.	1.6	31
126	Effects of aerobic training on exercise-related oxidative stress in mitochondrial myopathies. Neuromuscular Disorders, 2012, 22, S172-S177.	0.6	31

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127	Next-generation sequencing approach to hyperCKemia. Neurology: Genetics, 2019, 5, e352.	1.9	31
128	Visual hallucinations in Parkinson's disease are not influenced by polymorphisms of serotonin 5-HT2A receptor and transporter genes. Neuroscience Letters, 2007, 422, 228-231.	2.1	30
129	Bilateral striatal necrosis, dystonia and multiple mitochondrial DNA deletions: Case study and effect of deep brain stimulation. Movement Disorders, 2008, 23, 114-118.	3.9	30
130	Dysphagia in Amyotrophic Lateral Sclerosis: Relationships between disease progression and Fiberoptic Endoscopic Evaluation of Swallowing. Auris Nasus Larynx, 2017, 44, 306-312.	1.2	30
131	Impact of <scp>Coronavirus Disease 20</scp>19 Pandemic on Cognition in Parkinson's Disease. Movement Disorders, 2020, 35, 1717-1718.	3.9	30
132	The genetics of ataxia: through the labyrinth of the Minotaur, looking for Ariadneâ€™s thread. Journal of Neurology, 2014, 261, 528-541.	3.6	29
133	A longitudinal study of polysomnographic variables in patients with mild cognitive impairment converting to Alzheimer's disease. Journal of Sleep Research, 2019, 28, e12821.	3.2	29
134	Mitochondrial Syndromes Revisited. Journal of Clinical Medicine, 2021, 10, 1249.	2.4	29
135	Evaluation of cytogenetic and DNA damage in mitochondrial disease patients: effects of coenzyme Q10 therapy. Mutagenesis, 2004, 19, 43-49.	2.6	28
136	Epilepsy and limb girdle muscular dystrophy type 2A: double trouble, serendipitous finding or new phenotype?. Neurological Sciences, 2006, 27, 134-137.	1.9	28
137	MERRF syndrome without ragged-red fibers: The need for molecular diagnosis. Biochemical and Biophysical Research Communications, 2007, 354, 1058-1060.	2.1	28
138	Rippling muscle disease and facioscapulohumeral dystrophy-like phenotype in a patient carrying a heterozygous CAV3 T78M mutation and a D4Z4 partial deletion: Further evidence for â€œdouble troubleâ€• overlapping syndromes. Neuromuscular Disorders, 2012, 22, 534-540.	0.6	28
139	Alzheimerâ€™s Pathogenesis and Its Link to the Mitochondrion. Oxidative Medicine and Cellular Longevity, 2015, 2015, 1-8.	4.0	28
140	Fatigue, sleepâ€“wake pattern, depressive and anxiety symptoms and body-mass index: analysis in a sample of episodic and chronic migraine patients. Neurological Sciences, 2016, 37, 987-989.	1.9	28
141	Mitochondrial tRNACys gene mutation (A5814G): a second family with mitochondrial encephalopathy. Neuromuscular Disorders, 1997, 7, 156-159.	0.6	27
142	Missense and splice site mutations in SPG4 suggest loss-of-function in dominant spastic paraplegia. Journal of Neurology, 2002, 249, 200-205.	3.6	27
143	Autosomal dominant psychiatric disorders and mitochondrial DNA multiple deletions: Report of a family. Journal of Affective Disorders, 2008, 106, 173-177.	4.1	27
144	<scp>EFNS</scp> review on the role of muscle biopsy in the investigation of myalgia. European Journal of Neurology, 2013, 20, 997-1005.	3.3	27

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145	Cortical silent period in patients with amyotrophic lateral sclerosis. Journal of the Neurological Sciences, 1999, 169, 93-97.	0.6	25
146	Elevated TGF Î²2 serum levels in Emery-Dreifuss Muscular Dystrophy: Implications for myocyte and tenocyte differentiation and fibrogenic processes. Nucleus, 2018, 9, 337-349.	2.2	25
147	Pharmacogenetics of myotonic hNav1.4 sodium channel variants situated near the fast inactivation gate. Pharmacological Research, 2019, 141, 224-235.	7.1	25
148	Phenotypic Variability Among Patients With D4Z4 Reduced Allele Facioscapulohumeral Muscular Dystrophy. JAMA Network Open, 2020, 3, e204040.	5.9	25
149	Epidemiology of dystrophinopathies in North-West Tuscany: a molecular genetics-based revisitation. Clinical Genetics, 1999, 56, 51-58.	2.0	24
150	The relationship of plasma catecholamine and lactate during anaerobic threshold exercise in mitochondrial myopathies. Neuromuscular Disorders, 1999, 9, 411-416.	0.6	24
151	Detection of anti-ganglioside antibodies in Guillain-BarrÃ© syndrome and its variants by the agglutination assay. Journal of the Neurological Sciences, 2002, 196, 41-44.	0.6	24
152	Oxcarbazepine is effective and safe in the treatment of neuropathic pain: pooled analysis of seven clinical studies. Neurological Sciences, 2005, 26, 218-226.	1.9	24
153	D90A-SOD1 mutation in ALS: The first report of heterozygous Italian patients and unusual findings. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2010, 11, 216-219.	2.1	24
154	ClCâ€1 mutations in myotonia congenita patients: insights into molecular gating mechanisms and genotypeâ€“phenotype correlation. Journal of Physiology, 2015, 593, 4181-4199.	2.9	24
155	Multidisciplinary study of a new ClCâ€1 mutation causing myotonia congenita: a paradigm to understand and treat ion channelopathies. FASEB Journal, 2016, 30, 3285-3295.	0.5	24
156	Potential Diagnostic Value of Red Blood Cells Î±-Synuclein Heteroaggregates in Alzheimerâ€™s Disease. Molecular Neurobiology, 2019, 56, 6451-6459.	4.0	24
157	Estimating the impact of COVID-19 pandemic on services provided by Italian Neuromuscular Centers: an Italian Association of Myology survey of the acute phase. Acta Myologica, 2020, 39, 57-66.	1.5	24
158	Clock T3111C and Per2 C111G SNPs do not influence circadian rhythmicity in healthy Italian population. Neurological Sciences, 2011, 32, 89-93.	1.9	23
159	Data from the European registry for patients with McArdle disease and other muscle glycogenoses (EUROMAC). Orphanet Journal of Rare Diseases, 2020, 15, 330.	2.7	23
160	Anti-cN1A Antibodies Are Associated with More Severe Dysphagia in Sporadic Inclusion Body Myositis. Cells, 2021, 10, 1146.	4.1	23
161	Hereditary spastic paraparesis in adults. A clinical and genetic perspective from Tuscany. Clinical Neurology and Neurosurgery, 2014, 120, 14-19.	1.4	22
162	Frequency of diabetes and other comorbidities in chronic inflammatory demyelinating polyradiculoneuropathy and their impact on clinical presentation and response to therapy. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 1092-1099.	1.9	22

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163	Mapping Cortical Degeneration in ALS with Magnetization Transfer Ratio and Voxel-Based Morphometry. PLoS ONE, 2013, 8, e68279.	2.5	22
164	Altered surface myoelectric signals in peripheral vascular disease: Correlations with muscle fiber composition. , 1998, 21, 201-210.		21
165	Lack of Association between Nuclear Factor Erythroid-Derived 2-Like 2 Promoter Gene Polymorphisms and Oxidative Stress Biomarkers in Amyotrophic Lateral Sclerosis Patients. Oxidative Medicine and Cellular Longevity, 2014, 2014, 1-9.	4.0	21
166	Thyroid hormone levels in the cerebrospinal fluid correlate with disease severity in euthyroid patients with Alzheimerâ€™s disease. Endocrine, 2017, 55, 981-984.	2.3	21
167	A non-syndromic hearing loss caused by very low levels of the mtDNA A3243G mutation. Acta Neurologica Scandinavica, 2004, 110, 72-74.	2.1	20
168	Subclinical cardiac involvement in patients with facioscapulohumeral muscular dystrophy. Neuromuscular Disorders, 2005, 15, 403-408.	0.6	20
169	DPP6 gene variability confers increased risk of developing sporadic amyotrophic lateral sclerosis in Italian patients. Journal of Neurology, Neurosurgery and Psychiatry, 2008, 79, 1085-1085.	1.9	20
170	Vaccination recommendations for patients with neuromuscular disease. Vaccine, 2014, 32, 5893-5900.	3.8	20
171	The Methylenetetrahydrofolate Reductase C677T Polymorphism and Risk for Late-Onset Alzheimerâ€™s disease: Further Evidence in an Italian Multicenter Study. Journal of Alzheimer's Disease, 2017, 56, 1451-1457.	2.6	20
172	MRI cortical feature of bulbar impairment in patients with amyotrophic lateral sclerosis. NeuroImage: Clinical, 2019, 24, 101934.	2.7	20
173	Fluid Candidate Biomarkers for Alzheimerâ€™s Disease: A Precision Medicine Approach. Journal of Personalized Medicine, 2020, 10, 221.	2.5	20
174	Multicenter Study on Sleep and Circadian Alterations as Objective Markers of Mild Cognitive Impairment and Alzheimerâ€™s Disease Reveals Sex Differences. Journal of Alzheimer's Disease, 2020, 78, 1707-1719.	2.6	20
175	CSF sphingomyelin: a new biomarker of demyelination in the diagnosis and management of CIDP and GBS. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 303-310.	1.9	20
176	Muscle exercise in limb girdle muscular dystrophies: pitfall and advantages. Acta Myologica, 2015, 34, 3-8.	1.5	20
177	Magnetization Transfer Imaging Demonstrates a Distributed Pattern of Microstructural Changes of the Cerebral Cortex in Amyotrophic Lateral Sclerosis. American Journal of Neuroradiology, 2011, 32, 704-708.	2.4	19
178	Lipomatosis Incidence and Characteristics in an Italian Cohort of Mitochondrial Patients. Frontiers in Neurology, 2019, 10, 160.	2.4	19
179	Dopamine Transporter Imaging, Current Status of a Potential Biomarker: A Comprehensive Review. International Journal of Molecular Sciences, 2021, 22, 11234.	4.1	19
180	No major progranulin genetic variability contribution to disease etiopathogenesis in an ALS Italian cohort. Neurobiology of Aging, 2011, 32, 1157-1158.	3.1	18

#	Article	IF	Citations
181	Autonomic, functional, skeletal muscle, and cardiac abnormalities are associated with increased ergoreflex sensitivity in mitochondrial disease. European Journal of Heart Failure, 2017, 19, 1701-1709.	7.1	18
182	Î±-Synuclein Aggregated with Tau and Î²-Amyloid in Human Platelets from Healthy Subjects: Correlation with Physical Exercise. Frontiers in Aging Neuroscience, 2018, 10, 17.	3.4	18
183	Plasma Levels of Oxidative Stress Markers, before and after BoNT/A Treatment, in Chronic Migraine. Toxins, 2019, 11, 608.	3.4	18
184	Creutzfeldtâ€“Jakob disease with E200K PRNP mutation: a case report and revision of the literature. Neurological Sciences, 2009, 30, 417-420.	1.9	17
185	The hOGG1 Ser326Cys polymorphism is not associated with sporadic Parkinson's disease. Neuroscience Letters, 2010, 473, 248-251.	2.1	17
186	Transplanted Human Adipose Tissue-Derived Stem Cells Engraft and Induce Regeneration in Mice Olfactory Neuroepithelium in Response to Dichlobenil Subministration. Chemical Senses, 2014, 39, 617-629.	2.0	17
187	Novel POLG mutations and variable clinical phenotypes in 13 Italian patients. Neurological Sciences, 2017, 38, 563-570.	1.9	17
188	Semiautomated Evaluation of the Primary Motor Cortex in Patients with Amyotrophic Lateral Sclerosis at 3T. American Journal of Neuroradiology, 2018, 39, 63-69.	2.4	17
189	Telemedicine for neuromuscular disorders during the COVID-19 outbreak. Journal of Neurology, 2021, 268, 1-4.	3.6	17
190	Prolonged epileptic discharges predict seizure recurrence in JME: Insights from prolonged ambulatory EEG. Epilepsia, 2021, 62, 1184-1192.	5.1	17
191	International retrospective natural history study of LMNA-related congenital muscular dystrophy. Brain Communications, 2021, 3, fcab075.	3.3	17
192	Myophosphorylase deficiency affects muscle mitochondrial respiration as shown by 31P-MR spectroscopy in a case with associated multifocal encephalopathy. Journal of the Neurological Sciences, 1995, 128, 84-91.	0.6	16
193	Abnormal levels of human mitochondrial transcription factor A in skeletal muscle in mitochondrial encephalomyopathies. Neurological Sciences, 2000, 21, S985-S987.	1.9	16
194	Cardiac magnetic resonance imaging and management of dilated cardiomyopathy in a Duchenne muscular dystrophy manifesting carrier. Journal of Neurology, 2009, 256, 283-284.	3.6	16
195	Gly482Ser PGC-1Î± Gene Polymorphism and Exercise-Related Oxidative Stress in Amyotrophic Lateral Sclerosis Patients. Frontiers in Cellular Neuroscience, 2016, 10, 102.	3.7	16
196	Methylation analysis of DNA repair genes in Alzheimerâ€™s disease. Mechanisms of Ageing and Development, 2017, 161, 105-111.	4.6	16
197	A single center study: AÎ²42/p-Tau181 CSF ratio to discriminate AD from FTD in clinical setting. Neurological Sciences, 2017, 38, 1791-1797.	1.9	16
198	The role of synaptic biomarkers in the spectrum of neurodegenerative diseases. Expert Review of Proteomics, 2020, 17, 543-559.	3.0	16

#	Article	IF	Citations
199	Large genotypeâ€“phenotype study in carriers of D4Z4 borderline alleles provides guidance for facioscapulohumeral muscular dystrophy diagnosis. Scientific Reports, 2020, 10, 21648.	3.3	16
200	Respiratory chain defects in hereditary spastic paraplegias. Neuromuscular Disorders, 2001, 11, 565-569.	0.6	15
201	Novel spastin (SPG4) mutations in Italian patients with hereditary spastic paraplegia. Neuromuscular Disorders, 2006, 16, 387-390.	0.6	15
202	Mitochondria, oxidative stress and PARPâ€1 network: a new target for neuroprotective effects of tetracyclines?. Journal of Physiology, 2008, 586, 2427-2428.	2.9	15
203	Implication of a Genetic Variant at PICALM in Alzheimer's Disease Patients and Centenarians. Journal of Alzheimer's Disease, 2011, 24, 409-413.	2.6	15
204	A mobile app for patients with Pompe disease and its possible clinical applications. Neuromuscular Disorders, 2018, 28, 471-475.	0.6	15
205	Oxidative Stress Assessment in Alzheimerâ€™s Disease: A Clinic Setting Study. American Journal of Alzheimer's Disease and Other Dementias, 2018, 33, 35-41.	1.9	15
206	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
207	A 5-year clinical follow-up study from the Italian National Registry for FSHD. Journal of Neurology, 2021, 268, 356-366.	3.6	15
208	Identification of Serum Interleukin 6 Levels as a Disease Severity Biomarker in Facioscapulohumeral Muscular Dystrophy. Journal of Neuromuscular Diseases, 2022, 9, 83-93.	2.6	15
209	Myotonia permanens with Nav1.4-G1306E displays varied phenotypes during course of life. Acta Myologica, 2017, 36, 125-134.	1.5	15
210	Prevalent cardiac involvement in dystrophin Becker type mutation. Neuromuscular Disorders, 1994, 4, 381-386.	0.6	14
211	Residual muscle cytochrome c oxidase activity accounts for submaximal exercise lactate threshold in chronic progressive external ophthalmoplegia. , 1996, 19, 342-349.		14
212	Association study between XRCC1 gene polymorphisms and sporadic amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2010, 11, 122-124.	2.1	14
213	G41SSOD1mutation: A common ancestor for six ALS Italian families with an aggressive phenotype. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2010, 11, 210-215.	2.1	14
214	Protein misfolding, amyotrophic lateral sclerosis and guanabenz: protocol for a phase II RCT with futility design (ProMISe trial). BMJ Open, 2017, 7, e015434.	1.9	14
215	Disruption of sleep-wake continuum in myotonic dystrophy type 1: Beyond conventional sleep staging. Neuromuscular Disorders, 2018, 28, 414-421.	0.6	14
216	Mitochondrial epilepsy: a cross-sectional nationwide Italian survey. Neurogenetics, 2020, 21, 87-96.	1.4	14

#	Article	IF	Citations
217	Obstructive sleep apnea syndrome and Alzheimerâ€™s disease pathology: may continuous positive airway pressure treatment delay cognitive deterioration?. Sleep and Breathing, 2021, 25, 2135-2139.	1.7	14
218	Evoked Potentials in the Evaluation of Patients with Mitochondrial Myopathy. European Neurology, 1993, 33, 428-435.	1.4	13
219	The mtDNA A8344G â€œMERRFâ€•mutation is not a common cause of sporadic Parkinson disease in Italian population. Parkinsonism and Related Disorders, 2008, 14, 381-382.	2.2	13
220	A novel mitochondrial tRNAIle point mutation associated with chronic progressive external ophthalmoplegia and hyperCKemia. Journal of the Neurological Sciences, 2011, 300, 187-190.	0.6	13
221	Nerve and muscle involvement in mitochondrial disorders: an electrophysiological study. Neurological Sciences, 2012, 33, 449-452.	1.9	13
222	An â€œinflammatoryâ€•mitochondrial myopathy. A case report. Neuromuscular Disorders, 2013, 23, 907-910.	0.6	13
223	Evidences of Reduced Antioxidant Activity in Patients With Chronic Migraine and Medicationâ€Overuse Headache. Headache, 2015, 55, 984-991.	3.9	13
224	Next-generation sequencing application to investigate skeletal muscle channelopathies in a large cohort of Italian patients. Neuromuscular Disorders, 2021, 31, 336-347.	0.6	13
225	Cryoglobulinemic peripheral neuropathy: Neurophysiologic evaluation in twenty-two patients. Biomedicine and Pharmacotherapy, 1996, 50, 329-336.	5.6	12
226	The first Italian family with evidence of pyramidal impairment as phenotypic manifestation of Silver syndrome BSCL2 gene mutation. Neurological Sciences, 2008, 29, 189-191.	1.9	12
227	Selective techniques of apheresis in polyneuropathy associated with monoclonal gammopathy of undetermined significance. Acta Neurologica Scandinavica, 1994, 89, 117-122.	2.1	12
228	Lack of association between the APEX1 Asp148Glu polymorphism and sporadic amyotrophic lateral sclerosis. Neurobiology of Aging, 2010, 31, 353-355.	3.1	12
229	Prevalent cardiac phenotype resulting in heart transplantation in a novel LMNA gene duplication. Neuromuscular Disorders, 2010, 20, 512-516.	0.6	12
230	Phenotype may predict the clinical course of facioscapolohumeral muscular dystrophy. Muscle and Nerve, 2019, 59, 711-713.	2.2	12
231	Central Nervous System Involvement as Outcome Measure for Clinical Trials Efficacy in Myotonic Dystrophy Type 1. Frontiers in Neurology, 2020, 11, 624.	2.4	12
232	Oxidative stress biomarkers in Fabry disease: is there a room for them?. Journal of Neurology, 2020, 267, 3741-3752.	3.6	12
233	CGRP Inhibitors and Oxidative Stress Biomarkers in Resistant Migraine: A Real-Life Study with Erenumab, Fremanezumab, and Galcanezumab. Journal of Clinical Medicine, 2021, 10, 4586.	2.4	12
234	Non-radioactive detection of 17p11.2 duplication in CMT1A: a study of 78 patients.. Journal of Medical Genetics, 1994, 31, 880-883.	3.2	11

#	Article	IF	Citations
235	Facioscapulohumeral Muscular Dystrophy Type 1A in Northwestern Tuscany: A Molecular Genetics-based Epidemiological and Genotypeâ€“Phenotype Study. Genetic Testing and Molecular Biomarkers, 2005, 9, 30-36.	1.7	11
236	Mitochondrial DNA haplogroups do not influence the Huntington's disease phenotype. Neuroscience Letters, 2008, 444, 83-86.	2.1	11
237	Muscle metabolic alterations assessed by 31â€phosphorus magnetic resonance spectroscopy in mild becker muscular dystrophy. Muscle and Nerve, 2011, 44, 816-818.	2.2	11
238	A personal monitoring architecture to detect muscular fatigue in elderly. Neuromuscular Disorders, 2012, 22, S192-S197.	0.6	11
239	Cardiac involvement in chronic progressive external ophthalmoplegia. Journal of the Neurological Sciences, 2014, 345, 189-192.	0.6	11
240	Impact of ApoE Polymorphism and Physical Activity on Plasma Antioxidant Capability and Erythrocyte Membranes. Antioxidants, 2019, 8, 538.	5.1	11
241	Sleep Complaints, Sleep and Breathing Disorders in Myotonic Dystrophy Type 2. Current Neurology and Neuroscience Reports, 2019, 19, 9.	4.2	11
242	Cardiac magnetic resonance in patients with muscular dystrophies. European Journal of Preventive Cardiology, 2021, 28, 1526-1535.	1.8	11
243	Exercise-Related Oxidative Stress as Mechanism to Fight Physical Dysfunction in Neuromuscular Disorders. Frontiers in Physiology, 2020, 11, 451.	2.8	11
244	Apolipoprotein E Polymorphism and Oxidative Stress in Human Peripheral Blood Cells: Can Physical Activity Reactivate the Proteasome System through Epigenetic Mechanisms?. Oxidative Medicine and Cellular Longevity, 2021, 2021, 1-16.	4.0	11
245	Arterial intracranial thrombosis as the first manifestation of vaccine-induced immune thrombotic thrombocytopenia (VITT): a case report. Neurological Sciences, 2022, 43, 2085-2089.	1.9	11
246	Limbic encephalitis associated with thymic cancer: a case report. Journal of Neurology, 2001, 248, 1000-1002.	3.6	10
247	Tetracycline treatment in patients with progressive external ophthalmoplegia. Acta Neurologica Scandinavica, 2011, 124, 417-423.	2.1	10
248	Mitochondrial m.3243A>G mutation and carotid artery dissection. Molecular Genetics and Metabolism Reports, 2016, 9, 12-14.	1.1	10
249	Mitochondrial DNA haplogroups may influence Fabry disease phenotype. Neuroscience Letters, 2016, 629, 58-61.	2.1	10
250	Oxidative Stress in Cerebral Small Vessel Disease Dizziness Patients, Basally and After Polyphenol Compound Supplementation. Current Molecular Medicine, 2018, 18, 160-165.	1.3	10
251	Primary mitochondrial myopathy. Neurology: Genetics, 2020, 6, e519.	1.9	10
252	Clinical features of mtDNA-related syndromes in adulthood. Archives of Biochemistry and Biophysics, 2021, 697, 108689.	3.0	10

#	Article	IF	Citations
253	Diagnostic and prognostic value of CSF neurofilaments in a cohort of patients with motor neuron disease: A crossâ€sectional study. Journal of Cellular and Molecular Medicine, 2021, 25, 3765-3771.	3.6	10
254	Adult-onset mitochondrial movement disorders: a national picture from the Italian Network. Journal of Neurology, 2022, 269, 1413-1421.	3.6	10
255	Human mitochondrial transcription factor A reduction and mitochondrial dysfunction in Hashimoto's hypothyroid myopathy. Molecular Medicine, 2002, 8, 326-33.	4.4	10
256	Lithium in ALS: from the bench to the bedside. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2008, 9, 123-124.	2.1	9
257	Cognitive impairment and McArdle disease: Is there a link?. Neuromuscular Disorders, 2011, 21, 356-358.	0.6	9
258	Relation between plasmatic and cerebrospinal fluid oxidative stress biomarkers and intrathecal Ig synthesis in Multiple Sclerosis patients. Journal of Neuroimmunology, 2015, 283, 39-42.	2.3	9
259	Is early detection of late-onset Pompe disease a pneumologistâ€™s affair? A lesson from an Italian screening study. Orphanet Journal of Rare Diseases, 2019, 14, 62.	2.7	9
260	Fibroblast growth factor 21 and grow differentiation factor 15 are sensitive biomarkers of mitochondrial diseases due to mitochondrial transfer-RNA mutations and mitochondrial DNA deletions. Neurological Sciences, 2020, 41, 3653-3662.	1.9	9
261	Î±-Synuclein Heteromers in Red Blood Cells of Alzheimerâ€™s Disease and Lewy Body Dementia Patients. Journal of Alzheimer's Disease, 2021, 80, 885-893.	2.6	9
262	Increase in Mitochondrial D-Loop Region Methylation Levels in Mild Cognitive Impairment Individuals. International Journal of Molecular Sciences, 2022, 23, 5393.	4.1	9
263	Cytogenetic damage in peripheral lymphocytes of mitochondrial disease patients. Neurological Sciences, 2000, 21, S963-S965.	1.9	8
264	Effects of grating spatial orientation on visual evoked potentials and contrast sensitivity in multiple sclerosis. Acta Neurologica Scandinavica, 2001, 103, 97-104.	2.1	8
265	ELECTROPHYSIOLOGICAL EVALUATION OF GENITO-SPHINCTERIC DYSFUNCTION IN MULTIPLE SYSTEM ATROPHY. International Journal of Neuroscience, 2003, 113, 1353-1369.	1.6	8
266	Advanced oxidation protein products in serum of patients with myotonic disease type I: association with serum Î³-glutamyltransferase and disease severity. Clinical Chemistry and Laboratory Medicine, 2005, 43, 745-7.	2.3	8
267	Assessing the Role of Anti rh-GAA in Modulating Response to ERT in a Late-Onset Pompe Disease Cohort from the Italian GSDII Study Group. Advances in Therapy, 2019, 36, 1177-1189.	2.9	8
268	Psychosocial impact of sport activity in neuromuscular disorders. Neurological Sciences, 2020, 41, 2561-2567.	1.9	8
269	Progress regarding the context-of-use of tau as biomarker of Alzheimerâ€™s disease and other neurodegenerative diseases. Expert Review of Proteomics, 2021, 18, 27-48.	3.0	8
270	Movement Disorders in Children with a Mitochondrial Disease: A Cross-Sectional Survey from the Nationwide Italian Collaborative Network of Mitochondrial Diseases. Journal of Clinical Medicine, 2021, 10, 2063.	2.4	8

#	Article	IF	Citations
271	Statins in Parkinsonâ€™s Disease: Influence on Motor Progression. Journal of Parkinson's Disease, 2021, 11, 1651-1662.	2.8	8
272	Mutations associated with hypokalemic periodic paralysis: from hotspot regions to complete analysis of CACNA1S and SCN4A genes. Neurogenetics, 2022, 23, 19-25.	1.4	8
273	Dissection and atherosclerosis of carotid arteries in the young: role of the apolipoprotein E polymorphism. European Journal of Neurology, 2002, 9, 19-21.	3.3	7
274	Nerve, muscle and heart acute toxicity following oxaliplatin and capecitabine treatment. Neuromuscular Disorders, 2012, 22, 767-770.	0.6	7
275	The empowerment of translational research: lessons from laminopathies. Orphanet Journal of Rare Diseases, 2012, 7, 37.	2.7	7
276	Fatigue Is Increased in Episodic Migraine Without Aura Patients. Headache, 2013, 53, 1163-1165.	3.9	7
277	Levetiracetamâ€responsive myoclonus in spinocerebellar ataxia type 15. Movement Disorders, 2013, 28, 1465-1465.	3.9	7
278	Evaluation of damaging effects of splicing mutations: Validation of an in vitro method for diagnostic laboratories. Clinica Chimica Acta, 2014, 436, 276-282.	1.1	7
279	A novel family with axonal Charcotâ€Marieâ€Tooth disease caused by a mutation in the EGR2 gene. Journal of the Peripheral Nervous System, 2019, 24, 219-223.	3.1	7
280	Adapted physical activity and therapeutic exercise in late-onset Pompe disease (LOPD): a two-step rehabilitative approach. Neurological Sciences, 2020, 41, 859-868.	1.9	7
281	Clinical and Molecular Spectrum of Myotonia and Periodic Paralyses Associated With Mutations in SCN4A in a Large Cohort of Italian Patients. Frontiers in Neurology, 2020, 11, 646.	2.4	7
282	Increased resistance towards fatigability in patients with facioscapulohumeral muscular dystrophy. European Journal of Applied Physiology, 2021, 121, 1617-1629.	2.5	7
283	Expanding the clinical and genetic spectrum of pathogenic variants in <scp>STIM1</scp>. Muscle and Nerve, 2021, 64, 567-575.	2.2	7
284	Platelet-rich plasma: the role in neural repair. Neural Regeneration Research, 2015, 10, 1920.	3.0	7
285	Mitochondrial Ataxias: Molecular Classification and Clinical Heterogeneity. Neurology International, 2022, 14, 337-356.	2.8	7
286	Therapeutic opportunities and clinical outcome measures in Duchenne muscular dystrophy. Neurological Sciences, 2022, 43, 625-633.	1.9	7
287	Potential involvement of ubiquinone in myotonic dystrophy pathophysiology: new diagnostic approaches for new rationale therapeutics. Neurological Sciences, 2000, 21, S979-S980.	1.9	6
288	Sudden bilateral hearing loss and sporadic mitochondrial DNA deletion. Journal of Laryngology and Otology, 2001, 115, 128-131.	0.8	6

#	Article	IF	Citations
289	Molecular diagnostics and mitochondrial dysfunction: a future perspective. Expert Review of Molecular Diagnostics, 2008, 8, 531-549.	3.1	6
290	Novel MTCYB mutation in a young patient with recurrent strokeâ€like episodes and status epilepticus. American Journal of Medical Genetics, Part A, 2014, 164, 2922-2925.	1.2	6
291	Neurohormonal modulation for treatment of cardiac involvement in dystrophinopathies and mitochondrial disease. European Journal of Preventive Cardiology, 2017, 24, 1718-1724.	1.8	6
292	Supersaturation of VEP in Migraine without Aura Patients Treated with Topiramate: An Anatomo-Functional Biomarker of the Disease. Journal of Clinical Medicine, 2021, 10, 769.	2.4	6
293	Response to levetiracetam or lamotrigine in subjects with Juvenile Myoclonic Epilepsy previously treated with valproic acid: A single center retrospective study. Epilepsy and Behavior, 2021, 115, 107706.	1.7	6
294	A multi-parametric protocol to study exercise intolerance in McArdle's disease. Acta Myologica, 2015, 34, 120-125.	1.5	6
295	Mitochondrial ANT-1 related adPEO leading to cognitive impairment: is there a link?. Acta Myologica, 2017, 36, 25-27.	1.5	6
296	Exercise therapy in muscle diseases: open issues and future perspectives. Acta Myologica, 2019, 38, 233-238.	1.5	6
297	Association of rs3027178 polymorphism in the circadian clock gene PER1 with susceptibility to Alzheimerâ€™s disease and longevity in an Italian population. GeroScience, 2022, 44, 881-896.	4.6	6
298	Iron-sensitive MR imaging of the primary motor cortex to differentiate hereditary spastic paraplegia from other motor neuron diseases. European Radiology, 2022, 32, 8058-8064.	4.5	6
299	Ocular myopathy without ophthalmoplegia can be a form of mitochondrial myopathy. Clinical Neurology and Neurosurgery, 1992, 94, 133-141.	1.4	5
300	Variability of the expression of muscle mitochondrial damage in ocular mitochondrial myopathy. Neuromuscular Disorders, 1992, 2, 397-404.	0.6	5
301	Encephalomyopathy with multiple mitochondrial DNA deletions and multiple symmetric lipomatosis: further evidence of a possible association. Journal of Neurology, 1999, 246, 1197-1198.	3.6	5
302	Multiple mtDNA deletions: Clinical and molecular correlations. Journal of Inherited Metabolic Disease, 2000, 23, 155-161.	3.6	5
303	A novel out-of-frame mutation in the neurofilament light chain gene (NEFL) does not result in Charcot-Marie-Tooth disease type 2E. Neurogenetics, 2005, 6, 49-50.	1.4	5
304	Plastic Changes in the Spinal Cord in Motor Neuron Disease. BioMed Research International, 2014, 2014, 1-14.	1.9	5
305	Functional magnetic resonance imaging with encoding task in patients with mild cognitive impairment and different severity of leukoaraiosis. Psychiatry Research - Neuroimaging, 2018, 282, 126-131.	1.8	5
306	Positive DAT-SCAN in SPG7: a case report mimicking possible MSA-C. BMC Neurology, 2021, 21, 328.	1.8	5

#	Article	IF	Citations
307	Therapeutical Management and Drug Safety in Mitochondrial Diseasesâ€"Update 2020. Journal of Clinical Medicine, 2021, 10, 94.	2.4	5
308	The role of rehabilitation in the management of late-onset Pompe disease: a narrative review of the level of evidence. Acta Myologica, 2018, 37, 241-251.	1.5	5
309	Quality of Life in Adult Patients with Mitochondrial Myopathy. Neuroepidemiology, 2012, 38, 194-195.	2.3	5
310	Effects of Anticholinergic Agents on the Excitability of the Blink Reflex in Meige Syndrome. European Neurology, 1989, 29, 281-283.	1.4	4
311	A case of pelvic floor myoclonic jerk syndrome. Movement Disorders, 1996, 11, 331-333.	3.9	4
312	Mitochondrial DNA single deletion in a patient with postural tremor. Movement Disorders, 2008, 23, 2098-2100.	3.9	4
313	Fabry Disease With Atypical Neurological Presentation. Neurologist, 2012, 18, 413-414.	0.7	4
314	Inferior rectus myositis: a rare cause of painful ophthalmoplegia and a therapeutic challenge. Neurological Sciences, 2013, 34, 789-791.	1.9	4
315	Post-therapy normalization of brain FDG-PET in Morvan's syndrome. Journal of the Neurological Sciences, 2015, 353, 175-176.	0.6	4
316	Proximal Myopathy due to m.5835G>A Mutation in Mitochondrial MT-TY Gene. Case Reports in Neurological Medicine, 2018, 2018, 1-4.	0.4	4
317	Impact of environmental factors and physical activity on disability and quality of life in CIDP. Journal of Neurology, 2020, 267, 2683-2691.	3.6	4
318	Ultrasensitive techniques and protein misfolding amplification assays for biomarker-guided reconceptualization of Alzheimerâ€™s and other neurodegenerative diseases. Expert Review of Neurotherapeutics, 2021, 21, 949-967.	2.8	4
319	An updated review on the role of prescribed exercise in the management of Amyotrophic lateral sclerosis. Expert Review of Neurotherapeutics, 2021, 21, 871-879.	2.8	4
320	Fatigue as a common signature of inflammatory myopathies: clinical aspects and care.. Clinical and Experimental Rheumatology, 2022, 40, 425-432.	0.8	4
321	Fluid Biomarkers in Alzheimerâ€™s Disease and Other Neurodegenerative Disorders: Toward Integrative Diagnostic Frameworks and Tailored Treatments. Diagnostics, 2022, 12, 796.	2.6	4
322	Î±-synuclein as an emerging pathophysiological biomarker of Alzheimerâ€™s disease. Expert Review of Molecular Diagnostics, 2022, 22, 411-425.	3.1	4
323	Distribution Indices of Magnetic Susceptibility Values in the Primary Motor Cortex Enable to Classify Patients with Amyotrophic Lateral Sclerosis. Brain Sciences, 2022, 12, 942.	2.3	4
324	Effects of selenium and vitamin E on muscular strength and blood parameters in Steinert disease. Italian Journal of Neurological Sciences, 1990, 11, 37-42.	0.1	3

#	Article	IF	Citations
325	Is erythropoietin gene a modifier factor in amyotrophic lateral sclerosis?. Neurobiology of Aging, 2009, 30, 842-844.	3.1	3
326	A new truncating MPZ mutation associated with a very mild CMT1 B phenotype. Neuromuscular Disorders, 2010, 20, 817-819.	0.6	3
327	Acute encephalopathy of the temporal lobes leading to m.3243A > G. When MELAS is not always MELAS. Mitochondrion, 2016, 30, 148-150.	3.4	3
328	Plasma redox and inflammatory patterns during major depressive episodes: a cross-sectional investigation in elderly patients with mood disorders. CNS Spectrums, 2021, 26, 416-426.	1.2	3
329	Efficacy of fingolimod after switching from interferon Î²-1a in an adolescent with multiple sclerosis: case report. Neurological Sciences, 2021, 42, 5-7.	1.9	3
330	The neurophysiological lesson from the Italian CIDP database. Neurological Sciences, 2021, , 1.	1.9	3
331	SARS-CoV-2 infection in patients with primary mitochondrial diseases: Features and outcomes in Italy. Mitochondrion, 2021, 58, 243-245.	3.4	3
332	Prolonged and short epileptiform discharges have an opposite relationship with the sleepâ€“wake cycle in patients with JME: Implications for EEG recording protocols. Epilepsy and Behavior, 2021, 122, 108226.	1.7	3
333	Amyotrophic Lateral Sclerosis: A Genetic Point of View. Current Molecular Medicine, 2014, 14, 1089-1101.	1.3	3
334	Validation of the DYALS (dysphagia in amyotrophic lateral sclerosis) questionnaire for the evaluation of dysphagia in ALS patients. Neurological Sciences, 2022, 43, 3195-3200.	1.9	3
335	R1 responses of the trigeminofacial reflex in lesions extrinsic to the brain stem. Italian Journal of Neurological Sciences, 1984, 5, 41-44.	0.1	2
336	Toxic myopathy induced by industrial minerals oils: Clinical and histopathological features. Italian Journal of Neurological Sciences, 1986, 7, 599-604.	0.1	2
337	G.P.251. Neuromuscular Disorders, 2014, 24, 892.	0.6	2
338	Chronic inflammatory demyelinating polyradiculoneuropathy with cranial nerves hypertrophy, thyroid-related orbitopathy and IgG monoclonal gammopathy: a case report. Neurological Sciences, 2015, 36, 1027-1029.	1.9	2
339	Catatonia as prominent feature of stroke-like episode in MELAS. Neurological Sciences, 2021, 42, 383-385.	1.9	2
340	E-Health & Innovation to Overcome Barriers in Neuromuscular Diseases. Report from the 1st eNMD Congress: Nice, France, March 22-23, 2019. Journal of Neuromuscular Diseases, 2021, 8, 743-754.	2.6	2
341	A case of intravascular large B cell lymphoma with brain involvement mimicking progressive multifocal leukoencephalopathy. International Journal of Neuroscience, 2023, 133, 735-739.	1.6	2
342	Mitochondrial DNA (mtDNA) haplotypes and dysfunctions in presbyacusis. Acta Otorhinolaryngologica Italica, 2014, 34, 54-61.	1.5	2

#	Article	IF	Citations
343	Hard ways towards adulthood: the transition phase in young people with myotonic dystrophy. Acta Myologica, 2016, 35, 145-149.	1.5	2
344	Neuromuscular tetanic hyperexcitability syndrome associated to a heterozygous mutation with normal serum magnesium levels. Acta Myologica, 2020, 39, 36-39.	1.5	2
345	Ictogenesis of viral pneumonia: A comparison between SARS-CoV-2 and H1N1/H3N2. Epilepsy and Behavior, 2022, 126, 108470.	1.7	2
346	Mitochondrial stroke-like episodes: The search for new therapies. Pharmacological Research, 2022, 180, 106228.	7.1	2
347	A Single mtDNA Deletion in Association with a LMNA Gene New Frameshift Variant: A Case Report. Journal of Neuromuscular Diseases, 2022, 9, 457-462.	2.6	2
348	Reply to Vanacore and Galeotti: Sequential trials and the use of placebo. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, .	7.1	1
349	G.P.3.08 Myoclonic epilepsy, cortical dysplasia and mitochondrial complex I deficiency: A case report. Neuromuscular Disorders, 2009, 19, 563-564.	0.6	1
350	G.P.15.09 Unexpected high percentage of asymptomatic subjects carrying the FSHD molecular defect: Which factors contribute to the disease mechanism?. Neuromuscular Disorders, 2009, 19, 649-650.	0.6	1
351	Inflammatory myopathy in a patient with postural and kinetik tremor. Neurological Sciences, 2011, 32, 1175-1178.	1.9	1
352	Oxidative stress modulation in neurodegenerative diseases. Mediterranean Journal of Nutrition and Metabolism, 2011, 4, 219-225.	0.5	1
353	Editorial. Neuromuscular Disorders, 2012, 22, S155.	0.6	1
354	A Pilot Study Evaluating the Contribution ofSLC19A1(RFC-1) 80G›A Polymorphism to Alzheimerâ€™s Disease in Italian Caucasians. BioMed Research International, 2014, 2014, 1-6.	1.9	1
355	G.P.136. Neuromuscular Disorders, 2014, 24, 841-842.	0.6	1
356	P041. Analysis of body mass index, psychiatric comorbidity, sleep-wake pattern and occurrence of fatigue in episodic and chronic migraine patients. Journal of Headache and Pain, 2015, 16, A188.	6.0	1
357	Amyotrophic lateral sclerosis with long lasting disease course and SOD1 and TARDBP mutations: Report of two cases and overview of the literature. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2017, 18, 137-139.	1.7	1
358	CPEO and Mitochondrial Myopathy in a Patient with DGUOK Compound Heterozygous Pathogenetic Variant and mtDNA Multiple Deletions. Case Reports in Neurological Medicine, 2019, 2019, 1-4.	0.4	1
359	Prolonged distal motor latency of median nerve does not improve diagnostic accuracy for CIDP. Journal of Neurology, 2021, , 1.	3.6	1
360	Dissecting the Interplay Between Time of Dementia and Cognitive Profiles in Lewy Body Dementias. Journal of Alzheimer's Disease, 2021, 84, 757-766.	2.6	1

#	Article	IF	Citations
361	Anti-HMGCR antibodies and asymptomatic hyperCKemia. A case report. Acta Myologica, 2021, 40, 105-108.	1.5	1
362	Frailties and critical issues in neuromuscular diseases highlighted by SARS-CoV-2 pandemic: how many patients are still "invisible"?. Acta Myologica, 2022, 41, 24-29.	1.5	1
363	Is hypnotic assessment relevant to neurology?. Neurological Sciences, 2022, , 1.	1.9	1
364	Immunoglobulin treatment in myasthenia gravis. Electroencephalography and Clinical Neurophysiology, 1985, 61, S85.	0.3	0
365	The role of tactile pressure input in the habituation phenomenon of the trigemino-facial reflex. Electroencephalography and Clinical Neurophysiology, 1985, 61, S209.	0.3	0
366	Myokymic syndrome with impaired muscular relaxation: further evidence of a possible paraneoplastic genesis. Clinical Neurology and Neurosurgery, 1990, 92, 169-175.	1.4	0
367	A NOVEL MUTATION OF CONNEXINâ€32 GENE IN A CMTX ITALIAN FAMILY. Journal of the Peripheral Nervous System, 2000, 5, 51-51.	3.1	0
368	Clinical open-label study for evaluation of efficacy and tolerability of oxcarbazepine in the treatment of neuropathic pain. Journal of the Peripheral Nervous System, 2004, 9, 120-120.	3.1	0
369	G.P.7.02 Multicenter study on occurrence of auditory impairment in facioscapulohumeral muscular dystrophy. Neuromuscular Disorders, 2007, 17, 806.	0.6	0
370	G.P.14.04 Oxidative stress markers are reduced after cysteine donor enriched dietary intake in myotonic dystrophy type I. Neuromuscular Disorders, 2007, 17, 855.	0.6	0
371	D.P.1.02 A robust tool to quantify disability in patients affected by facioscapulohumeral muscular dystrophy. Neuromuscular Disorders, 2008, 18, 726-727.	0.6	0
372	Reply to Bedlack et al. : A small pilot study calls for large clinical trials to evaluate the effects of lithium before prescribing the drug for amyotrophic lateral sclerosis. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, .	7.1	0
373	G.P.3.10 Oxidative stress biomarkers are related to disease severity in mitochondrial myopathies, and may be modified by a cysteine donor. Neuromuscular Disorders, 2009, 19, 564.	0.6	0
374	G.P.15.08 High genetic variability in European population: The FSHD complex puzzle. Neuromuscular Disorders, 2009, 19, 649.	0.6	0
375	P1.19 Whole genetic and protein characterisation in DMD symptomatic female carriers excludes correlation with X-inactivation and transcriptional DMD allele balancing. Neuromuscular Disorders, 2011, 21, 647.	0.6	0
376	Oxidative stress modulation in neurodegenerative diseases. Mediterranean Journal of Nutrition and Metabolism, 2011, 4, 219-225.	0.5	0
377	Muscle fatigue in neuromuscular disorders: Pathogenic mechanisms and treatment (following a) Tj ETQq1 1 0.784314 rgBT /Overlock	0.6	0
378	Reply to the letter. Neuromuscular Disorders, 2013, 23, 525.	0.6	0

#	Article	IF	Citations
379	P.5.17 LMNA-associated myopathies: The Italian Laminopathies Network experience in a large cohort of patients. Neuromuscular Disorders, 2013, 23, 769.	0.6	0
380	Differential effects of alendronate and losartan therapy on osteopenia and aortic aneurysm in mice with severe Marfan syndrome. Human Molecular Genetics, 2014, 23, 6137-6137.	2.9	0
381	G.O.7. Neuromuscular Disorders, 2014, 24, 851.	0.6	0
382	Clinical and molecular features of a large cohort of Italian McArdle patients. Neuromuscular Disorders, 2015, 25, S219.	0.6	0
383	Large screening of patients diagnosed as limb girdle muscular dystrophy or congenital myopathy using Motorplex. Neuromuscular Disorders, 2015, 25, S297.	0.6	0
384	Biomarkers and progress of antioxidant therapy for rare mitochondrial disorders. Expert Opinion on Orphan Drugs, 2016, 4, 591-603.	0.8	0
385	Red blood cell Î±â€synuclein heteroaggregates can discriminate healthy controls from cognitively impaired subjects of the ADâ€LBD spectrum. Alzheimer's and Dementia, 2020, 16, e040618.	0.8	0
386	Sex differences in red blood cell Î± â€synuclein protein and its heteroaggregates with amyloidâ€Î² and tau in early Alzheimerâ€™s disease. Alzheimer's and Dementia, 2020, 16, e042079.	0.8	0
387	Assessment of the integrity of the noradrenergic nucleus locus coeruleus during normal ageing by neuromelaninâ€3T MRI. Alzheimer's and Dementia, 2020, 16, e043332.	0.8	0
388	In vivo assessment of the noradrenergic nucleus locus coeruleus in Alzheimerâ€™s disease and other types of dementia. Alzheimer's and Dementia, 2020, 16, e043616.	0.8	0
389	Cardiac magnetic resonance findings in patients with type 1 myotonic dystrophy. European Heart Journal Cardiovascular Imaging, 2021, 22, .	1.2	0
390	Muscle Fiber Conduction Velocity Correlates With the Age at Onset in Mild FSHD Cases. Frontiers in Physiology, 2021, 12, 686176.	2.8	0
391	Response to â€œSalvage therapy for vagal nerve stimulator infection; Literature review and report of a delayed recurrenceâ€• Clinical Neurology and Neurosurgery, 2021, 207, 106721.	1.4	0
392	Unusual concomitant smallâ€•and largeâ€fiber neuropathy related to hypereosinophilic syndrome. Clinical and Experimental Neuroimmunology, 0, , .	1.0	0
393	31â€ƒCardiac magnetic resonance findings in patients with Type 1 myotonic dystrophy. European Heart Journal Supplements, 2021, 23, .	0.1	0
394	Fatigue as a common signature of inflammatory myopathies: clinical aspects and care.. Clinical and Experimental Rheumatology, 2022, 40, 425-432.	0.8	0