Nereo Bresolin

List of Publications by Year
in descending order

408 papers	18,295 citations	11608 70 h-index	24915 109 g-index
412 all docs	412 docs citations	412 times ranked	24819 citing authors

#	Article	IF	Citations
1	Aging-Dependent Large Accumulation of Point Mutations in the Human mtDNA Control Region for Replication. Science, 1999, 286, 774-779.	6.0	691
2	Cell Therapy of Â-Sarcoglycan Null Dystrophic Mice Through Intra-Arterial Delivery of Mesoangioblasts. Science, 2003, 301, 487-492.	6.0	593
3	Disorders of cognitive and affective development in cerebellar malformations. Brain, 2007, 130, 2646-2660.	3.7	324
4	Human circulating AC133+ stem cells restore dystrophin expression and ameliorate function in dystrophic skeletal muscle. Journal of Clinical Investigation, 2004, 114, 182-195.	3.9	315
5	Intrathecal Chemokine Synthesis in Mild Cognitive Impairment and Alzheimer Disease. Archives of Neurology, 2006, 63, 538.	4.9	268
6	Cytochromec Oxidase subunit I microdeletion in a patient with motor neuron disease. Annals of Neurology, 1998, 43, 110-116.	2.8	251
7	Intraarterial Injection of Muscle-Derived Cd34+Sca-1+ Stem Cells Restores Dystrophin in mdx Mice. Journal of Cell Biology, 2001, 152, 335-348.	2.3	248
8	Identification of a Primitive Brain-Derived Neural Stem Cell Population Based on Aldehyde Dehydrogenase Activity. Stem Cells, 2006, 24, 975-985.	1.4	240
9	Parasites represent a major selective force for interleukin genes and shape the genetic predisposition to autoimmune conditions. Journal of Experimental Medicine, 2009, 206, 1395-1408.	4.2	230
10	Restoration of Human Dystrophin Following Transplantation of Exon-Skipping-Engineered DMD Patient Stem Cells into Dystrophic Mice. Cell Stem Cell, 2007, 1, 646-657.	5.2	206
11	Serum MCP-1 levels are increased in mild cognitive impairment and mild Alzheimer's disease. Neurobiology of Aging, 2006, 27, 1763-1768.	1.5	185
12	Genetic Correction of Human Induced Pluripotent Stem Cells from Patients with Spinal Muscular Atrophy. Science Translational Medicine, 2012, 4, 165ra162.	5.8	180
13	The Role of Mitochondria in Neurodegenerative Diseases: the Lesson from Alzheimerâ€™s Disease and Parkinsonâ€™s Disease. Molecular Neurobiology, 2020, 57, 2959-2980.	1.9	180
14	Oxidative imbalance in patients with mild cognitive impairment and Alzheimer's disease. Neurobiology of Aging, 2006, 27, 262-269.	1.5	178
15	Selective DNA Methylation of BDNF Promoter in Bipolar Disorder: Differences Among Patients with BDI and BDII. Neuropsychopharmacology, 2012, 37, 1647-1655.	2.8	166
16	Clinical, molecular, and protein correlations in a large sample of genetically diagnosed Italian limb girdle muscular dystrophy patients. Human Mutation, 2008, 29, 258-266.	1.1	162
17	Muscle Coenzyme Q10 Level in Statin-Related Myopathy. Archives of Neurology, 2005, 62, 1709.	4.9	158
18	Cognitive impairment in Duchenne muscular dystrophy. Neuromuscular Disorders, 1994, 4, 359-369.	0.3	152

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19	Widespread balancing selection and pathogen-driven selection at blood group antigen genes. Genome Research, 2009, 19, 199-212.	2.4	147
20	Expression and genetic analysis of miRNAs involved in CD4+ cell activation in patients with multiple sclerosis. Neuroscience Letters, 2011, 504, 9-12.	1.0	147
21	Direct reprogramming of human astrocytes into neural stem cells and neurons. Experimental Cell Research, 2012, 318, 1528-1541.	1.2	143
22	Intracellular Delivery of a Tat-eGFP Fusion Protein into Muscle Cells. Molecular Therapy, 2001, 3, 310-318.	3.7	139
23	Genotype and phenotype characterization in a large dystrophinopathic cohort with extended follow-up. Journal of Neurology, 2011, 258, 1610-1623.	1.8	134
24	Agenesis of the Corpus Callosum: Clinical and Genetic Study in 63 Young Patients. Pediatric Neurology, 2006, 34, 186-193.	1.0	130
25	Human skin-derived stem cells migrate throughout forebrain and differentiate into astrocytes after injection into adult mouse brain. Journal of Neuroscience Research, 2004, 77, 475-486.	1.3	129
26	Neural stem cells LewisX + CXCR4 + modify disease progression in an amyotrophic lateral sclerosis model. Brain, 2007, 130, 1289-1305.	3.7	127
27	?-enolase deficiency, a new metabolic myopathy of distal glycolysis. Annals of Neurology, 2001, 50, 202-207.	2.8	125
28	Mitochondrial Respiratory Chain Dysfunction in Muscle From Patients With Amyotrophic Lateral Sclerosis. Archives of Neurology, 2010, 67, 849-54.	4.9	122
29	The Mitochondrial Disulfide Relay System Protein GFER Is Mutated in Autosomal-Recessive Myopathy with Cataract and Combined Respiratory-Chain Deficiency. American Journal of Human Genetics, 2009, 84, 594-604.	2.6	121
30	Silencer elements as possible inhibitors of pseudoexon splicing. Nucleic Acids Research, 2004, 32, 1783-1791.	6.5	120
31	Neural stem cell transplantation can ameliorate the phenotype of a mouse model of spinal muscular atrophy. Journal of Clinical Investigation, 2008, 118, 3316-3330.	3.9	119
32	Stem cell therapy in stroke. Cellular and Molecular Life Sciences, 2009, 66, 757-772.	2.4	119
33	Vascular endothelial growth factor gene variability is associated with increased risk for AD. Annals of Neurology, 2005, 57, 373-380.	2.8	115
34	Mutations in DNA2 Link Progressive Myopathy to Mitochondrial DNA Instability. American Journal of Human Genetics, 2013, 92, 293-300.	2.6	115
35	MFN2-related neuropathies: Clinical features, molecular pathogenesis and therapeutic perspectives. Journal of the Neurological Sciences, 2015, 356, 7-18.	0.3	112
36	Neural Stem Cell Transplantation for Neurodegenerative Diseases. International Journal of Molecular Sciences, 2020, 21, 3103.	1.8	105

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37	Production of monocyte chemoattractant protein-1 in amyotrophic lateral sclerosis. Muscle and Nerve, 2005, 32, 541-544.	1.0	104
38	Isolation and characterization of murine neural stem/progenitor cells based on Prominin-1 expression. Experimental Neurology, 2007, 205, 547-562.	2.0	104
39	Marked agingâ€related decline in efficiency of oxidative phosphorylation in human skin fibroblasts. FASEB Journal, 2003, 17, 1706-1708.	0.2	102
40	Retrospective study of a large population of patients with asymptomatic or minimally symptomatic raised serum creatine kinase levels. Journal of Neurology, 2002, 249, 305-311.	1.8	100
41	POLG mutations in sporadic mitochondrial disorders with multiple mtDNA deletions. Human Mutation, 2003, 22, 498-499.	1.1	100
42	Gait pattern in Duchenne muscular dystrophy. Gait and Posture, 2009, 29, 36-41.	0.6	99
43	Embryonic stem cell-derived neural stem cells improve spinal muscular atrophy phenotype in mice. Brain, 2010, 133, 465-481.	3.7	98
44	Decreased circulating miRNA levels in patients with primary progressive multiple sclerosis. Multiple Sclerosis Journal, 2013, 19, 1938-1942.	1.4	98
45	Minimally invasive transplantation of iPSC-derived ALDHhiSSCloVLA4+ neural stem cells effectively improves the phenotype of an amyotrophic lateral sclerosis model. Human Molecular Genetics, 2014, 23, 342-354.	1.4	97
46	Progressive cytochromec oxidase deficiency in a case of earns-sayre syndrome: Morphological, immunological, and biochemical studies in muscle biopsies and autopsy tissues. Annals of Neurology, 1987, 21, 564-572.	2.8	96
47	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.	1.7	93
48	Transplanted ALDHhiSSClo neural stem cells generate motor neurons and delay disease progression of nmd mice, an animal model of SMARD1. Human Molecular Genetics, 2006, 15, 167-187.	1.4	90
49	Muscle mitochondrial DNA deletion and 31P-NMR spectroscopy alterations in a migraine patient. Journal of the Neurological Sciences, 1991, 104, 182-189.	0.3	89
50	Parkinson's Disease and Brain Mitochondrial Dysfunction: A Functional Phosphorus Magnetic Resonance Spectroscopy Study. Journal of Cerebral Blood Flow and Metabolism, 2006, 26, 283-290.	2.4	89
51	The first ALS2 missense mutation associated with JPLS reveals new aspects of alsin biological function. Brain, 2006, 129, 1710-1719.	3.7	87
52	Spontaneous blinking in healthy persons: an optoelectronic study of eyelid motion. Ophthalmic and Physiological Optics, 2008, 28, 345-353.	1.0	87
53	Aging-dependent Functional Alterations of Mitochondrial DNA (mtDNA) from Human Fibroblasts Transferred into mtDNA-less Cells. Journal of Biological Chemistry, 1996, 271, 15891-15897.	1.6	86
54	Neuroectodermal and microglial differentiation of bone marrow cells in the mouse spinal cord and sensory ganglia. Journal of Neuroscience Research, 2002, 70, 721-733.	1.3	86

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55	The italian limb girdle muscular dystrophy registry: Relative frequency, clinical features, and differential diagnosis. Muscle and Nerve, 2017, 55, 55-68.	1.0	86
56	Carnitine, carnitine acyltransferases, and rat brain function. Experimental Neurology, 1982, 78, 285-292.	2.0	85
57	A clinical, genetic, and biochemical characterization ofSPG7mutations in a large cohort of patients with hereditary spastic paraplegia. Human Mutation, 2008, 29, 522-531.	1.1	85
58	Mitochondrial Fusion Proteins and Human Diseases. Neurology Research International, 2013, 2013, 1-11.	0.5	85
59	Genetic diversity at endoplasmic reticulum aminopeptidases is maintained by balancing selection and is associated with natural resistance to HIV-1 infection. Human Molecular Genetics, 2010, 19, 4705-4714.	1.4	84
60	Progranulin plasma levels as potential biomarker for the identification of GRN deletion carriers. A case with atypical onset as clinical amnestic Mild Cognitive Impairment converted to Alzheimer's disease. Journal of the Neurological Sciences, 2009, 287, 291-293.	0.3	83
61	Abdominal volume contribution to tidal volume as an early indicator of respiratory impairment in Duchenne muscular dystrophy. European Respiratory Journal, 2010, 35, 1118-1125.	3.1	82
62	Growth factors in ischemic stroke. Journal of Cellular and Molecular Medicine, 2011, 15, 1645-1687.	1.6	81
63	Next-generation sequencing reveals DGUOK mutations in adult patients with mitochondrial DNA multiple deletions. Brain, 2012, 135, 3404-3415.	3.7	81
64	Multisystem triglyceride storage disorder with impaired long-chain fatty acid oxidation. Annals of Neurology, 1980, 7, 5-10.	2.8	79
65	MCP-1 in Alzheimerâ€™s disease patients: A-2518G polymorphism and serum levels. Neurobiology of Aging, 2004, 25, 1169-1173.	1.5	77
66	Intrathecal levels of IL-6, IL-11 and LIF in Alzheimer's disease and frontotemporal lobar degeneration. Journal of Neurology, 2008, 255, 539-544.	1.8	76
67	The apolipoprotein E Ïµ4 allele causes a faster decline of cognitive performances in Down's syndrome subjects. Journal of the Neurological Sciences, 1997, 145, 87-91.	0.3	75
68	Cognitive impairment in neuromuscular disorders. Muscle and Nerve, 2006, 34, 16-33.	1.0	74
69	Defective autophagy in spastizin mutated patients with hereditary spastic paraparesis type 15. Brain, 2013, 136, 3119-3139.	3.7	74
70	Molecular Therapeutic Strategies for Spinal Muscular Atrophies: Current and Future Clinical Trials. Clinical Therapeutics, 2014, 36, 128-140.	1.1	74
71	A Subpopulation of Murine Bone Marrow Cells Fully Differentiates along the Myogenic Pathway and Participates in Muscle Repair in the mdx Dystrophic Mouse. Experimental Cell Research, 2002, 277, 74-85.	1.2	70
72	Genome-Wide Identification of Susceptibility Alleles for Viral Infections through a Population Genetics Approach. PLoS Genetics, 2010, 6, e1000849.	1.5	67

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73	Loss of Dp140 regulatory sequences is associated with cognitive impairment in dystrophinopathies. Neuromuscular Disorders, 2000, 10, 194-199.	0.3	66
74	Analysis of intronic conserved elements indicates that functional complexity might represent a major source of negative selection on non-coding sequences. Human Molecular Genetics, 2005, 14, 2533-2546.	1.4	66
75	Systemic transplantation of c-kit+ cells exerts a therapeutic effect in a model of amyotrophic lateral sclerosis. Human Molecular Genetics, 2010, 19, 3782-3796.	1.4	66
76	Multipotentiality, homing properties, and pyramidal neurogenesis of CNSâ€derived LeX(sseaâ€1) + /CXCR4 + stem cells. FASEB Journal, 2005, 19, 1860-1862.	0.2	65
77	A New Mitochondrial DNA Mutation in ND3 Gene Causing Severe Leigh Syndrome with Early Lethality. Pediatric Research, 2004, 55, 842-846.	1.1	64
78	New Mutations in TK2 Gene Associated With Mitochondrial DNA Depletion. Pediatric Neurology, 2006, 34, 177-185.	1.0	63
79	Inclusion body myopathy and frontotemporal dementia caused by a novel VCP mutation. Neurobiology of Aging, 2009, 30, 752-758.	1.5	63
80	Phenotypic Heterogeneity of the GRN Asp22fs Mutation in a Large Italian Kindred. Journal of Alzheimer's Disease, 2011, 24, 253-259.	1.2	62
81	Mutations in CYP2U1, DDHD2 and GBA2 genes are rare causes of complicated forms of hereditary spastic paraparesis. Journal of Neurology, 2014, 261, 373-381.	1.8	62
82	Generation of skeletal muscle cells from embryonic and induced pluripotent stem cells as an in vitro model and for therapy of muscular dystrophies. Journal of Cellular and Molecular Medicine, 2012, 16, 1353-1364.	1.6	61
83	The signature of long-standing balancing selection at the human defensin Î²-1 promoter. Genome Biology, 2008, 9, R143.	13.9	60
84	Early Onset Behavioral Variant Frontotemporal Dementia due to the C9ORF72 Hexanucleotide Repeat Expansion: Psychiatric Clinical Presentations. Journal of Alzheimer's Disease, 2012, 31, 447-452.	1.2	60
85	Identification of a putative pathway for the muscle homing of stem cells in a muscular dystrophy model. Journal of Cell Biology, 2003, 162, 511-520.	2.3	59
86	Rs5848 Variant Influences GRN mRNA Levels in Brain and Peripheral Mononuclear Cells in Patients with Alzheimer's Disease. Journal of Alzheimer's Disease, 2009, 18, 603-612.	1.2	59
87	The landscape of human genes involved in the immune response to parasitic worms. BMC Evolutionary Biology, 2010, 10, 264.	3.2	59
88	CHCHD10mutations in Italian patients with sporadic amyotrophic lateral sclerosis: Figure 1. Brain, 2015, 138, e372-e372.	3.7	59
89	Population Genetics of IFIH1: Ancient Population Structure, Local Selection, and Implications for Susceptibility to Type 1 Diabetes. Molecular Biology and Evolution, 2010, 27, 2555-2566.	3.5	58
90	Brain Activation in Normal Subjects and in Patients Affected by Mitochondrial Disease without Clinical Central Nervous System Involvement: A Phosphorus Magnetic Resonance Spectroscopy Study. Journal of Cerebral Blood Flow and Metabolism, 2001, 21, 85-91.	2.4	57

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91	Role of hnRNP-A1 and miR-590-3p in Neuronal Death: Genetics and Expression Analysis in Patients with Alzheimer Disease and Frontotemporal Lobar Degeneration. Rejuvenation Research, 2011, 14, 275-281.	0.9	57
92	OASes and STING: Adaptive Evolution in Concert. Genome Biology and Evolution, 2015, 7, 1016-1032.	1.1	57
93	High tumor necrosis factor-? in levels in cerebrospinal fluid of cobalamin-deficient patients. Annals of Neurology, 2004, 56, 886-890.	2.8	55
94	Effect of Human Skin-Derived Stem Cells on Vessel Architecture, Tumor Growth, and Tumor Invasion in Brain Tumor Animal Models. Cancer Research, 2007, 67, 3054-3063.	0.4	55
95	The Low-Affinity Receptor for Neurotrophins p75^{NTR} Plays a Key Role for Satellite Cell Function in Muscle Repair Acting via RhoA. Molecular Biology of the Cell, 2009, 20, 3620-3627.	0.9	55
96	Variants in KIF1A gene in dominant and sporadic forms of hereditary spastic paraparesis. Journal of Neurology, 2015, 262, 2684-2690.	1.8	55
97	The evolutionary history of genes involved in spoken and written language: beyond FOXP2. Scientific Reports, 2016, 6, 22157.	1.6	55
98	Is M129V of PRNP gene associated with Alzheimer's disease? A case-control study and a meta-analysis. Neurobiology of Aging, 2006, 27, 770.e1-770.e5.	1.5	54
99	A novelCLN8mutation in late-infantile-onset neuronal ceroid lipofuscinosis (LINCL) reveals aspects of CLN8 neurobiological function. Human Mutation, 2009, 30, 1104-1116.	1.1	53
100	Frequency and characterisation of anoctamin 5 mutations in a cohort of Italian limb-girdle muscular dystrophy patients. Neuromuscular Disorders, 2012, 22, 934-943.	0.3	53
101	Ongoing therapeutic trials and outcome measures for Duchenne muscular dystrophy. Cellular and Molecular Life Sciences, 2013, 70, 4585-4602.	2.4	53
102	Brain Mitochondria, Aging, and Parkinsonâ€™s Disease. Genes, 2018, 9, 250.	1.0	53
103	MicroRNA expression analysis identifies a subset of downregulated miRNAs in ALS motor neuron progenitors. Scientific Reports, 2018, 8, 10105.	1.6	53
104	Chemotactic Factors Enhance Myogenic Cell Migration across an Endothelial Monolayer. Experimental Cell Research, 2001, 268, 36-44.	1.2	52
105	Quantitative muscle strength assessment in duchenne muscular dystrophy: longitudinal study and correlation with functional measures. BMC Neurology, 2012, 12, 91.	0.8	52
106	A wide spectrum of clinical, neurophysiological and neuroradiological abnormalities in a family with a novel CACNA1A mutation. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, 840-843.	0.9	51
107	Beta-lactam antibiotic offers neuroprotection in a spinal muscular atrophy model by multiple mechanisms. Experimental Neurology, 2011, 229, 214-225.	2.0	51
108	Both selective and neutral processes drive GC content evolution in the human genome. BMC Evolutionary Biology, 2008, 8, 99.	3.2	50

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109	A novel mutation in the Î²â€tubulin gene TUBB2B associated with complex malformation of cortical development and deficits in axonal guidance. Developmental Medicine and Child Neurology, 2012, 54, 765-769.	1.1	50
110	â€œEars of the Lynxâ€•MRI Sign Is Associated with SPG11 and SPG15 Hereditary Spastic Paraplegia. American Journal of Neuroradiology, 2019, 40, 199-203.	1.2	50
111	Silence superoxide dismutase 1 (SOD1): a promising therapeutic target for amyotrophic lateral sclerosis (ALS). Expert Opinion on Therapeutic Targets, 2020, 24, 295-310.	1.5	49
112	Intragenic Inversion of mtDNA: A New Type of Pathogenic Mutation in a Patient with Mitochondrial Myopathy. American Journal of Human Genetics, 2000, 66, 1900-1904.	2.6	48
113	Skeletal muscle gene expression profiling in mitochondrial disorders. FASEB Journal, 2005, 19, 1-30.	0.2	48
114	Antisense Oligonucleotide Therapy for the Treatment of C9ORF72 ALS/FTD Diseases. Molecular Neurobiology, 2014, 50, 721-732.	1.9	48
115	Fas small interfering RNA reduces motoneuron death in amyotrophic lateral sclerosis mice. Annals of Neurology, 2007, 62, 81-92.	2.8	47
116	Diagnostic and Prognostic Role of Blood and Cerebrospinal Fluid and Blood Neurofilaments in Amyotrophic Lateral Sclerosis: A Review of the Literature. International Journal of Molecular Sciences, 2019, 20, 4152.	1.8	47
117	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.	1.6	47
118	Clinical features and new molecular findings in Carnitine Palmitoyltransferase II (CPT II) deficiency. Journal of the Neurological Sciences, 2008, 266, 97-103.	0.3	46
119	Neurocognitive Profiles in Duchenne Muscular Dystrophy and Gene Mutation Site. Pediatric Neurology, 2011, 45, 292-299.	1.0	46
120	Assessing mental health in boys with Duchenne muscular dystrophy: Emotional, behavioural and neurodevelopmental profile in an Italian clinical sample. European Journal of Paediatric Neurology, 2017, 21, 639-647.	0.7	46
121	Mitochondrial Dysregulation and Impaired Autophagy in iPSC-Derived Dopaminergic Neurons of Multiple System Atrophy. Stem Cell Reports, 2018, 11, 1185-1198.	2.3	46
122	A Functional Variant in ERAP1 Predisposes to Multiple Sclerosis. PLoS ONE, 2012, 7, e29931.	1.1	46
123	Retrospective study of a large population of patients affected with mitochondrial disorders: clinical, morphological and molecular genetic evaluation. Journal of Neurology, 2001, 248, 778-788.	1.8	45
124	Molecular analysis of LGMD-2B and MM patients: identification of novel DYSF mutations and possible founder effect in the Italian population. Neuromuscular Disorders, 2003, 13, 788-795.	0.3	45
125	Novel Twinkle (PEO1) gene mutations in mendelian progressive external ophthalmoplegia. Journal of Neurology, 2008, 255, 1384-1391.	1.8	45
126	Cerebrospinal Fluid Biomarkers in Progranulin Mutations Carriers. Journal of Alzheimer's Disease, 2011, 27, 781-790.	1.2	45

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127	Effect of Combined Systemic and Local Morpholino Treatment on the Spinal Muscular Atrophy Î”7 Mouse Model Phenotype. Clinical Therapeutics, 2014, 36, 340-356.e5.	1.1	44
128	Focal cognitive impairment in mitochondrial encephalomyopathies: a neuropsychological and neuroimaging study. Journal of the Neurological Sciences, 1999, 170, 57-63.	0.3	43
129	Cryptogenic Epileptic Syndromes Related to SCN1A. Archives of Neurology, 2008, 65, 489.	4.9	43
130	Evidence and age-related distribution of mtDNA D-loop point mutations in skeletal muscle from healthy subjects and mitochondrial patients. Journal of the Neurological Sciences, 2002, 202, 85-91.	0.3	42
131	An Evolutionary Analysis of Antigen Processing and Presentation across Different Timescales Reveals Pervasive Selection. PLoS Genetics, 2014, 10, e1004189.	1.5	42
132	Subclinical leukodystrophy and infertility in a man with a novel homozygous CLCN2 mutation. Neurology, 2014, 83, 1217-1218.	1.5	42
133	Extracellular vesicles and amyotrophic lateral sclerosis: from misfolded protein vehicles to promising clinical biomarkers. Cellular and Molecular Life Sciences, 2021, 78, 561-572.	2.4	42
134	Absence of angiogenic genes modification in Italian ALS patients. Neurobiology of Aging, 2008, 29, 314-316.	1.5	41
135	Clinical and Paraclinical Indicators of Motor System Impairment in Hereditary Spastic Paraplegia: A Pilot Study. PLoS ONE, 2016, 11, e0153283.	1.1	41
136	Selective mitochondrial depletion, apoptosis resistance, and increased mitophagy in human Charcot-Marie-Tooth 2A motor neurons. Human Molecular Genetics, 2016, 25, 4266-4281.	1.4	41
137	Multiple deletions of mitochondrial DNA in sporadic and atypical cases of encephalomyopathy. Journal of the Neurological Sciences, 1994, 123, 74-79.	0.3	40
138	High mutational burden in the mtDNA control region from aged muscles: a single-fiber study. Neurobiology of Aging, 2003, 24, 829-838.	1.5	40
139	Intron size in mammals: complexity comes to terms with economy. Trends in Genetics, 2007, 23, 20-24.	2.9	40
140	Human Fetal Brain Chemistry as Detected by Proton Magnetic Resonance Spectroscopy. Pediatric Neurology, 2009, 40, 327-342.	1.0	40
141	Mitochondrial DNA G8363A mutation in the tRNALys gene: Clinical, biochemical and pathological study. Journal of the Neurological Sciences, 2009, 281, 85-92.	0.3	40
142	Nitric oxide donor and non steroidal anti inflammatory drugs as a therapy for muscular dystrophies: Evidence from a safety study with pilot efficacy measures in adult dystrophic patients. Pharmacological Research, 2012, 65, 472-479.	3.1	40
143	Early onset, non fluctuating spinocerebellar ataxia and a novel missense mutation in CACNA1A gene. Journal of the Neurological Sciences, 2006, 241, 13-17.	0.3	39
144	Induced neural stem cells: Methods of reprogramming and potential therapeutic applications. Progress in Neurobiology, 2014, 114, 15-24.	2.8	39

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145	R-Loops in Motor Neuron Diseases. Molecular Neurobiology, 2019, 56, 2579-2589.	1.9	39
146	Congenital myopathy associated with abnormal accumulation of desmin and dystrophin. Neuromuscular Disorders, 1992, 2, 169-175.	0.3	38
147	SPG11: a consistent clinical phenotype in a family with homozygous Spatacsin truncating mutation. Neurogenetics, 2007, 8, 301-305.	0.7	38
148	Spinal muscular atrophy phenotype is ameliorated in human motor neurons by SMN increase via different novel RNA therapeutic approaches. Scientific Reports, 2015, 5, 11746.	1.6	37
149	Multiparametric quantitative MRI assessment of thigh muscles in limbâ€girdle muscular dystrophy 2A and 2B. Muscle and Nerve, 2018, 58, 550-558.	1.0	37
150	Muscle glucose-6-phosphate dehydrogenase deficiency. Journal of Neurology, 1989, 236, 193-198.	1.8	36
151	Influence of the Glu298Asp polymorphism of NOS3 on age at onset and homocysteine levels in AD patients. Neurobiology of Aging, 2005, 26, 789-794.	1.5	36
152	Preconditioning and Cellular Engineering to Increase the Survival of Transplanted Neural Stem Cells for Motor Neuron Disease Therapy. Molecular Neurobiology, 2019, 56, 3356-3367.	1.9	36
153	CCR2-64I polymorphism and CCR5Î”32 deletion in patients with Alzheimer's disease. Journal of the Neurological Sciences, 2004, 225, 79-83.	0.3	35
154	A novel de novo nonsense mutation in ATP1A2 associated with sporadic hemiplegic migraine and epileptic seizures. Journal of the Neurological Sciences, 2008, 273, 123-126.	0.3	35
155	Amyotrophic lateral sclerosis linked to a novel SOD1 mutation with muscle mitochondrial dysfunction. Journal of the Neurological Sciences, 2009, 276, 170-174.	0.3	35
156	Respiratory pattern in an adult population of dystrophic patients. Journal of the Neurological Sciences, 2011, 306, 54-61.	0.3	35
157	Glial cells involvement in spinal muscular atrophy: Could SMA be a neuroinflammatory disease?. Neurobiology of Disease, 2020, 140, 104870.	2.1	35
158	Acute diazepam administration produces rapid increases in brain benzodiazepine receptor density. European Journal of Pharmacology, 1979, 59, 159-160.	1.7	34
159	VEGF gene variability and type 1 diabetes: evidence for a protective role. Immunogenetics, 2006, 58, 107-112.	1.2	34
160	Motoneuron Transplantation Rescues the Phenotype of SMARD1 (Spinal Muscular Atrophy with) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 1	1.7	34
161	Diverse Evolutionary Histories for Î²-adrenoreceptor Genes in Humans. American Journal of Human Genetics, 2009, 85, 64-75.	2.6	34
162	VEGF genetic variability is associated with increased risk of developing Alzheimer's disease. Journal of the Neurological Sciences, 2009, 283, 66-68.	0.3	34

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163	Progranulin Gene Variability and Plasma Levels in Bipolar Disorder and Schizophrenia. PLoS ONE, 2012, 7, e32164.	1.1	34
164	iPSC-Derived Neural Stem Cells Act via Kinase Inhibition to Exert Neuroprotective Effects in Spinal Muscular Atrophy with Respiratory DistressÂType 1. Stem Cell Reports, 2014, 3, 297-311.	2.3	34
165	Management of patients with neuromuscular disorders at the time of the SARS-CoV-2 pandemic. Journal of Neurology, 2021, 268, 1580-1591.	1.8	34
166	Primary beta-sarcoglycanopathy manifesting as recurrent exercise-induced myoglobinuria. Neuromuscular Disorders, 2001, 11, 389-394.	0.3	33
167	The dystrophin gene is alternatively spliced throughout its coding sequence. FEBS Letters, 2002, 517, 163-166.	1.3	33
168	Gene therapy rescues disease phenotype in a spinal muscular atrophy with respiratory distress type 1 (SMARD1) mouse model. Science Advances, 2015, 1, e1500078.	4.7	33
169	Novel missense mutation and large deletion ofGNE gene in autosomal-recessive inclusion-body myopathy. Muscle and Nerve, 2003, 28, 113-117.	1.0	32
170	Lactate detection in the brain of growth-restricted fetuses with magnetic resonance spectroscopy. American Journal of Obstetrics and Gynecology, 2011, 205, 350.e1-350.e7.	0.7	32
171	Clinical and molecular characterization of a cohort of patients with novel nucleotide alterations of the Dystrophin gene detected by direct sequencing. BMC Medical Genetics, 2011, 12, 37.	2.1	32
172	Stem cell transplantation for amyotrophic lateral sclerosis: therapeutic potential and perspectives on clinical translation. Cellular and Molecular Life Sciences, 2014, 71, 3257-3268.	2.4	32
173	Mitochondrial dysfunction in fibroblasts of Multiple System Atrophy. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 3588-3597.	1.8	32
174	Human induced pluripotent stem cell models for the study and treatment of Duchenne and Becker muscular dystrophies. Therapeutic Advances in Neurological Disorders, 2019, 12, 175628641983347.	1.5	32
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