Nereo Bresolin

List of Publications by Year
in descending order

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408	18,295	11651	24982
papers	citations	70 h-index	109 g-index

412	412	412	24819
all docs	docs citations	times ranked	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.	12.6	691
2	Cell Therapy of Â-Sarcoglycan Null Dystrophic Mice Through Intra-Arterial Delivery of Mesoangioblasts. Science, 2003, 301, 487-492.	12.6	593
3	Disorders of cognitive and affective development in cerebellar malformations. Brain, 2007, 130, 2646-2660.	7.6	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.	8.2	315
5	Intrathecal Chemokine Synthesis in Mild Cognitive Impairment and Alzheimer Disease. Archives of Neurology, 2006, 63, 538.	4.5	268
6	Cytochrome c Oxidase subunit I microdeletion in a patient with motor neuron disease. Annals of Neurology, 1998, 43, 110-116.	5.3	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.	5.2	248
8	Identification of a Primitive Brainâ€“Derived Neural Stem Cell Population Based on Aldehyde Dehydrogenase Activity. Stem Cells, 2006, 24, 975-985.	3.2	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.	8.5	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.	11.1	206
11	Serum MCP-1 levels are increased in mild cognitive impairment and mild Alzheimer's disease. Neurobiology of Aging, 2006, 27, 1763-1768.	3.1	185
12	Genetic Correction of Human Induced Pluripotent Stem Cells from Patients with Spinal Muscular Atrophy. Science Translational Medicine, 2012, 4, 165ra162.	12.4	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.	4.0	180
14	Oxidative imbalance in patients with mild cognitive impairment and Alzheimer's disease. Neurobiology of Aging, 2006, 27, 262-269.	3.1	178
15	Selective DNA Methylation of BDNF Promoter in Bipolar Disorder: Differences Among Patients with BDI and BDII. Neuropsychopharmacology, 2012, 37, 1647-1655.	5.4	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.	2.5	162
17	Muscle Coenzyme Q10 Level in Statin-Related Myopathy. Archives of Neurology, 2005, 62, 1709.	4.5	158
18	Cognitive impairment in Duchenne muscular dystrophy. Neuromuscular Disorders, 1994, 4, 359-369.	0.6	152

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19	Widespread balancing selection and pathogen-driven selection at blood group antigen genes. Genome Research, 2009, 19, 199-212.	5.5	147
20	Expression and genetic analysis of miRNAs involved in CD4+ cell activation in patients with multiple sclerosis. Neuroscience Letters, 2011, 504, 9-12.	2.1	147
21	Direct reprogramming of human astrocytes into neural stem cells and neurons. Experimental Cell Research, 2012, 318, 1528-1541.	2.6	143
22	Intracellular Delivery of a Tat-eGFP Fusion Protein into Muscle Cells. Molecular Therapy, 2001, 3, 310-318.	8.2	139
23	Genotype and phenotype characterization in a large dystrophinopathic cohort with extended follow-up. Journal of Neurology, 2011, 258, 1610-1623.	3.6	134
24	Agenesis of the Corpus Callosum: Clinical and Genetic Study in 63 Young Patients. Pediatric Neurology, 2006, 34, 186-193.	2.1	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.	2.9	129
26	Neural stem cells LewisX + CXCR4 + modify disease progression in an amyotrophic lateral sclerosis model. Brain, 2007, 130, 1289-1305.	7.6	127
27	?-enolase deficiency, a new metabolic myopathy of distal glycolysis. Annals of Neurology, 2001, 50, 202-207.	5.3	125
28	Mitochondrial Respiratory Chain Dysfunction in Muscle From Patients With Amyotrophic Lateral Sclerosis. Archives of Neurology, 2010, 67, 849-54.	4.5	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.	6.2	121
30	Silencer elements as possible inhibitors of pseudoexon splicing. Nucleic Acids Research, 2004, 32, 1783-1791.	14.5	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.	8.2	119
32	Stem cell therapy in stroke. Cellular and Molecular Life Sciences, 2009, 66, 757-772.	5.4	119
33	Vascular endothelial growth factor gene variability is associated with increased risk for AD. Annals of Neurology, 2005, 57, 373-380.	5.3	115
34	Mutations in DNA2 Link Progressive Myopathy to Mitochondrial DNA Instability. American Journal of Human Genetics, 2013, 92, 293-300.	6.2	115
35	MFN2-related neuropathies: Clinical features, molecular pathogenesis and therapeutic perspectives. Journal of the Neurological Sciences, 2015, 356, 7-18.	0.6	112
36	Neural Stem Cell Transplantation for Neurodegenerative Diseases. International Journal of Molecular Sciences, 2020, 21, 3103.	4.1	105

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37	Production of monocyte chemoattractant proteinâ€1 in amyotrophic lateral sclerosis. Muscle and Nerve, 2005, 32, 541-544.	2.2	104
38	Isolation and characterization of murine neural stem/progenitor cells based on Prominin-1 expression. Experimental Neurology, 2007, 205, 547-562.	4.1	104
39	Marked agingâ€related decline in efficiency of oxidative phosphorylation in human skin fibroblasts. FASEB Journal, 2003, 17, 1706-1708.	0.5	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.	3.6	100
41	POLG mutations in sporadic mitochondrial disorders with multiple mtDNA deletions. Human Mutation, 2003, 22, 498-499.	2.5	100
42	Gait pattern in Duchenne muscular dystrophy. Gait and Posture, 2009, 29, 36-41.	1.4	99
43	Embryonic stem cell-derived neural stem cells improve spinal muscular atrophy phenotype in mice. Brain, 2010, 133, 465-481.	7.6	98
44	Decreased circulating miRNA levels in patients with primary progressive multiple sclerosis. Multiple Sclerosis Journal, 2013, 19, 1938-1942.	3.0	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.	2.9	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.	5.3	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.	3.3	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.	2.9	90
49	Muscle mitochondrial DNA deletion and 31P-NMR spectroscopy alterations in a migraine patient. Journal of the Neurological Sciences, 1991, 104, 182-189.	0.6	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.	4.3	89
51	The first ALS2 missense mutation associated with JPLS reveals new aspects of alsin biological function. Brain, 2006, 129, 1710-1719.	7.6	87
52	Spontaneous blinking in healthy persons: an optoelectronic study of eyelid motion. Ophthalmic and Physiological Optics, 2008, 28, 345-353.	2.0	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.	3.4	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.	2.9	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.	2.2	86
56	Carnitine, carnitine acyltransferases, and rat brain function. Experimental Neurology, 1982, 78, 285-292.	4.1	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.	2.5	85
58	Mitochondrial Fusion Proteins and Human Diseases. Neurology Research International, 2013, 2013, 1-11.	1.3	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.	2.9	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.	0.6	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.	6.7	82
62	Growth factors in ischemic stroke. Journal of Cellular and Molecular Medicine, 2011, 15, 1645-1687.	3.6	81
63	Next-generation sequencing reveals DGUOK mutations in adult patients with mitochondrial DNA multiple deletions. Brain, 2012, 135, 3404-3415.	7.6	81
64	Multisystem triglyceride storage disorder with impaired longâ€chain fatty acid oxidation. Annals of Neurology, 1980, 7, 5-10.	5.3	79
65	MCP-1 in Alzheimerâ€™s disease patients: A-2518G polymorphism and serum levels. Neurobiology of Aging, 2004, 25, 1169-1173.	3.1	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.	3.6	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.	0.6	75
68	Cognitive impairment in neuromuscular disorders. Muscle and Nerve, 2006, 34, 16-33.	2.2	74
69	Defective autophagy in spastizin mutated patients with hereditary spastic paraparesis type 15. Brain, 2013, 136, 3119-3139.	7.6	74
70	Molecular Therapeutic Strategies for Spinal Muscular Atrophies: Current and Future Clinical Trials. Clinical Therapeutics, 2014, 36, 128-140.	2.5	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.	2.6	70
72	Genome-Wide Identification of Susceptibility Alleles for Viral Infections through a Population Genetics Approach. PLoS Genetics, 2010, 6, e1000849.	3.5	67

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73	Loss of Dp140 regulatory sequences is associated with cognitive impairment in dystrophinopathies. Neuromuscular Disorders, 2000, 10, 194-199.	0.6	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.	2.9	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.	2.9	66
76	Multipotentiality, homing properties, and pyramidal neurogenesis of CNSâ€derived LeX(sseaâ€1) + /CXCR4 + stem cells. FASEB Journal, 2005, 19, 1860-1862.	0.5	65
77	A New Mitochondrial DNA Mutation in ND3 Gene Causing Severe Leigh Syndrome with Early Lethality. Pediatric Research, 2004, 55, 842-846.	2.3	64
78	New Mutations in TK2 Gene Associated With Mitochondrial DNA Depletion. Pediatric Neurology, 2006, 34, 177-185.	2.1	63
79	Inclusion body myopathy and frontotemporal dementia caused by a novel VCP mutation. Neurobiology of Aging, 2009, 30, 752-758.	3.1	63
80	Phenotypic Heterogeneity of the GRN Asp22fs Mutation in a Large Italian Kindred. Journal of Alzheimer's Disease, 2011, 24, 253-259.	2.6	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.	3.6	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.	3.6	61
83	The signature of long-standing balancing selection at the human defensin Î²-1 promoter. Genome Biology, 2008, 9, R143.	9.6	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.	2.6	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.	5.2	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.	2.6	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.	7.6	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.	8.9	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.	4.3	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.	1.8	57
92	OASes and STING: Adaptive Evolution in Concert. Genome Biology and Evolution, 2015, 7, 1016-1032.	2.5	57
93	High tumor necrosis factor-? in levels in cerebrospinal fluid of cobalamin-deficient patients. Annals of Neurology, 2004, 56, 886-890.	5.3	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.	0.9	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.	2.1	55
96	Variants in KIF1A gene in dominant and sporadic forms of hereditary spastic paraparesis. Journal of Neurology, 2015, 262, 2684-2690.	3.6	55
97	The evolutionary history of genes involved in spoken and written language: beyond FOXP2. Scientific Reports, 2016, 6, 22157.	3.3	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.	3.1	54
99	A novelCLN8mutation in late-infantile-onset neuronal ceroid lipofuscinosis (LINCL) reveals aspects of CLN8 neurobiological function. Human Mutation, 2009, 30, 1104-1116.	2.5	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.	0.6	53
101	Ongoing therapeutic trials and outcome measures for Duchenne muscular dystrophy. Cellular and Molecular Life Sciences, 2013, 70, 4585-4602.	5.4	53
102	Brain Mitochondria, Aging, and Parkinsonâ€™s Disease. Genes, 2018, 9, 250.	2.4	53
103	MicroRNA expression analysis identifies a subset of downregulated miRNAs in ALS motor neuron progenitors. Scientific Reports, 2018, 8, 10105.	3.3	53
104	Chemotactic Factors Enhance Myogenic Cell Migration across an Endothelial Monolayer. Experimental Cell Research, 2001, 268, 36-44.	2.6	52
105	Quantitative muscle strength assessment in duchenne muscular dystrophy: longitudinal study and correlation with functional measures. BMC Neurology, 2012, 12, 91.	1.8	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.	1.9	51
107	Beta-lactam antibiotic offers neuroprotection in a spinal muscular atrophy model by multiple mechanisms. Experimental Neurology, 2011, 229, 214-225.	4.1	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.	2.1	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.	2.4	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.	3.4	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.	6.2	48
113	Skeletal muscle gene expression profiling in mitochondrial disorders. FASEB Journal, 2005, 19, 1-30.	0.5	48
114	Antisense Oligonucleotide Therapy for the Treatment of C9ORF72 ALS/FTD Diseases. Molecular Neurobiology, 2014, 50, 721-732.	4.0	48
115	Fas small interfering RNA reduces motoneuron death in amyotrophic lateral sclerosis mice. Annals of Neurology, 2007, 62, 81-92.	5.3	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.	4.1	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.	3.6	47
118	Clinical features and new molecular findings in Carnitine Palmitoyltransferase II (CPT II) deficiency. Journal of the Neurological Sciences, 2008, 266, 97-103.	0.6	46
119	Neurocognitive Profiles in Duchenne Muscular Dystrophy and Gene Mutation Site. Pediatric Neurology, 2011, 45, 292-299.	2.1	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.	1.6	46
121	Mitochondrial Dysregulation and Impaired Autophagy in iPSC-Derived Dopaminergic Neurons of Multiple System Atrophy. Stem Cell Reports, 2018, 11, 1185-1198.	4.8	46
122	A Functional Variant in ERAP1 Predisposes to Multiple Sclerosis. PLoS ONE, 2012, 7, e29931.	2.5	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.	3.6	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.	0.6	45
125	Novel Twinkle (PEO1) gene mutations in mendelian progressive external ophthalmoplegia. Journal of Neurology, 2008, 255, 1384-1391.	3.6	45
126	Cerebrospinal Fluid Biomarkers in Progranulin Mutations Carriers. Journal of Alzheimer's Disease, 2011, 27, 781-790.	2.6	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.	2.5	44
128	Focal cognitive impairment in mitochondrial encephalomyopathies: a neuropsychological and neuroimaging study. Journal of the Neurological Sciences, 1999, 170, 57-63.	0.6	43
129	Cryptogenic Epileptic Syndromes Related to SCN1A. Archives of Neurology, 2008, 65, 489.	4.5	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.	0.6	42
131	An Evolutionary Analysis of Antigen Processing and Presentation across Different Timescales Reveals Pervasive Selection. PLoS Genetics, 2014, 10, e1004189.	3.5	42
132	Subclinical leukodystrophy and infertility in a man with a novel homozygous CLCN2 mutation. Neurology, 2014, 83, 1217-1218.	1.1	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.	5.4	42
134	Absence of angiogenic genes modification in Italian ALS patients. Neurobiology of Aging, 2008, 29, 314-316.	3.1	41
135	Clinical and Paraclinical Indicators of Motor System Impairment in Hereditary Spastic Paraplegia: A Pilot Study. PLoS ONE, 2016, 11, e0153283.	2.5	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.	2.9	41
137	Multiple deletions of mitochondrial DNA in sporadic and atypical cases of encephalomyopathy. Journal of the Neurological Sciences, 1994, 123, 74-79.	0.6	40
138	High mutational burden in the mtDNA control region from aged muscles: a single-fiber study. Neurobiology of Aging, 2003, 24, 829-838.	3.1	40
139	Intron size in mammals: complexity comes to terms with economy. Trends in Genetics, 2007, 23, 20-24.	6.7	40
140	Human Fetal Brain Chemistry as Detected by Proton Magnetic Resonance Spectroscopy. Pediatric Neurology, 2009, 40, 327-342.	2.1	40
141	Mitochondrial DNA G8363A mutation in the tRNALys gene: Clinical, biochemical and pathological study. Journal of the Neurological Sciences, 2009, 281, 85-92.	0.6	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.	7.1	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.	0.6	39
144	Induced neural stem cells: Methods of reprogramming and potential therapeutic applications. Progress in Neurobiology, 2014, 114, 15-24.	5.7	39

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145	R-Loops in Motor Neuron Diseases. Molecular Neurobiology, 2019, 56, 2579-2589.	4.0	39
146	Congenital myopathy associated with abnormal accumulation of desmin and dystrophin. Neuromuscular Disorders, 1992, 2, 169-175.	0.6	38
147	SPG11: a consistent clinical phenotype in a family with homozygous Spatacsin truncating mutation. Neurogenetics, 2007, 8, 301-305.	1.4	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.	3.3	37
149	Multiparametric quantitative MRI assessment of thigh muscles in limbâ€girdle muscular dystrophy 2A and 2B. Muscle and Nerve, 2018, 58, 550-558.	2.2	37
150	Muscle glucose-6-phosphate dehydrogenase deficiency. Journal of Neurology, 1989, 236, 193-198.	3.6	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.	3.1	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.	4.0	36
153	CCR2-64I polymorphism and CCR5Î”32 deletion in patients with Alzheimer's disease. Journal of the Neurological Sciences, 2004, 225, 79-83.	0.6	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.	0.6	35
155	Amyotrophic lateral sclerosis linked to a novel SOD1 mutation with muscle mitochondrial dysfunction. Journal of the Neurological Sciences, 2009, 276, 170-174.	0.6	35
156	Respiratory pattern in an adult population of dystrophic patients. Journal of the Neurological Sciences, 2011, 306, 54-61.	0.6	35
157	Glial cells involvement in spinal muscular atrophy: Could SMA be a neuroinflammatory disease?. Neurobiology of Disease, 2020, 140, 104870.	4.4	35
158	Acute diazepam administration produces rapid increases in brain benzodiazepine receptor density. European Journal of Pharmacology, 1979, 59, 159-160.	3.5	34
159	VEGF gene variability and type 1 diabetes: evidence for a protective role. Immunogenetics, 2006, 58, 107-112.	2.4	34
160	Motoneuron Transplantation Rescues the Phenotype of SMARD1 (Spinal Muscular Atrophy with) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 1	3.6	34
161	Diverse Evolutionary Histories for Î²-adrenoreceptor Genes in Humans. American Journal of Human Genetics, 2009, 85, 64-75.	6.2	34
162	VEGF genetic variability is associated with increased risk of developing Alzheimer's disease. Journal of the Neurological Sciences, 2009, 283, 66-68.	0.6	34

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163	Progranulin Gene Variability and Plasma Levels in Bipolar Disorder and Schizophrenia. PLoS ONE, 2012, 7, e32164.	2.5	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.	4.8	34
165	Management of patients with neuromuscular disorders at the time of the SARS-CoV-2 pandemic. Journal of Neurology, 2021, 268, 1580-1591.	3.6	34
166	Primary beta-sarcoglycanopathy manifesting as recurrent exercise-induced myoglobinuria. Neuromuscular Disorders, 2001, 11, 389-394.	0.6	33
167	The dystrophin gene is alternatively spliced throughout its coding sequence. FEBS Letters, 2002, 517, 163-166.	2.8	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.	10.3	33
169	Novel missense mutation and large deletion ofGNE gene in autosomal-recessive inclusion-body myopathy. Muscle and Nerve, 2003, 28, 113-117.	2.2	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.	1.3	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.	5.4	32
173	Mitochondrial dysfunction in fibroblasts of Multiple System Atrophy. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 3588-3597.	3.8	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.	3.5	32
175	Association of a NOS1 promoter repeat with Alzheimer's disease. Neurobiology of Aging, 2008, 29, 1359-1365.	3.1	31
176	Role of OLR1 and Its Regulating hsa-miR369-3p in Alzheimer's Disease: Genetics and Expression Analysis. Journal of Alzheimer's Disease, 2011, 26, 787-793.	2.6	31
177	A POSITIVELY SELECTED APOBEC3H HAPLOTYPE IS ASSOCIATED WITH NATURAL RESISTANCE TO HIV-1 INFECTION. Evolution; International Journal of Organic Evolution, 2011, 65, 3311-3322.	2.3	31
178	Key role of SMN/SYNCRIP and RNA-Motif 7 in spinal muscular atrophy: RNA-Seq and motif analysis of human motor neurons. Brain, 2019, 142, 276-294.	7.6	31
179	Inducible nitric oxide synthase (iNOS) in immune-mediated demyelination and Wallerian degeneration of the rat peripheral nervous system. Experimental Neurology, 2004, 187, 350-358.	4.1	30
180	Gender-specific influence of the chromosome 16 chemokine gene cluster on the susceptibility to Multiple Sclerosis. Journal of the Neurological Sciences, 2008, 267, 86-90.	0.6	30

#	Article	IF	Citations
181	The role of protozoa-driven selection in shaping human genetic variability. Trends in Genetics, 2010, 26, 95-99.	6.7	30
182	Progranulin gene (GRN) promoter methylation is increased in patients with sporadic frontotemporal lobar degeneration. Neurological Sciences, 2013, 34, 899-903.	1.9	30
183	A 175 Million Year History of T Cell Regulatory Molecules Reveals Widespread Selection, with Adaptive Evolution of Disease Alleles. Immunity, 2013, 38, 1129-1141.	14.3	30
184	Development of Therapeutics for C9ORF72 ALS/FTD-Related Disorders. Molecular Neurobiology, 2017, 54, 4466-4476.	4.0	30
185	The 129 codon polymorphism of the Prion Protein gene influences earlier cognitive performance in Down syndrome subjects. Journal of Neurology, 2003, 250, 688-692.	3.6	29
186	Effects of rituximab in two patients with dysferlin-deficient muscular dystrophy. BMC Musculoskeletal Disorders, 2010, 11, 157.	1.9	29
187	Familial mtDNA T8993C transition causing both the NARP and the MILS phenotype in the same generation. Journal of Neurology, 2003, 250, 1498-1500.	3.6	28
188	Fixation of conserved sequences shapes human intron size and influences transposon-insertion dynamics. Trends in Genetics, 2005, 21, 484-488.	6.7	28
189	Gene function and expression level influence the insertion/fixation dynamics of distinct transposon families in mammalian introns. Genome Biology, 2006, 7, R120.	9.6	28
190	GRN Variability Contributes to Sporadic Frontotemporal Lobar Degeneration. Journal of Alzheimer's Disease, 2010, 19, 171-177.	2.6	28
191	A Novel MAPT Mutation Associated with the Clinical Phenotype of Progressive Nonfluent Aphasia. Journal of Alzheimer's Disease, 2011, 26, 19-26.	2.6	28
192	Senataxin modulates neurite growth through fibroblast growth factor 8 signalling. Brain, 2011, 134, 1808-1828.	7.6	28
193	Crohn's Disease Loci Are Common Targets of Protozoa-Driven Selection. Molecular Biology and Evolution, 2013, 30, 1077-1087.	8.9	28
194	Genetic adaptation of the human circadian clock to day-length latitudinal variations and relevance for affective disorders. Genome Biology, 2014, 15, 499.	8.8	28
195	Ancient and Recent Selective Pressures Shaped Genetic Diversity at AIM2-Like Nucleic Acid Sensors. Genome Biology and Evolution, 2014, 6, 830-845.	2.5	28
196	Glycogen storage disease type III: A novel Agl knockout mouse model. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2014, 1842, 2318-2328.	3.8	28
197	A novel mitochondrial tRNA Ile point mutation in chronic progressive external ophthalmoplegia. Journal of Neurology, 1998, 245, 755-758.	3.6	27
198	Absence of brain Dp140 isoform and cognitive impairment in Becker muscular dystrophy. Lancet, The, 1999, 353, 897-898.	13.7	27

#	Article	IF	Citations
199	Incontinence in Late-Onset Pompe Disease: An Underdiagnosed Treatable Condition. European Neurology, 2012, 68, 75-78.	1.4	27
200	A Trans-Specific Polymorphism in ZC3HAV1 Is Maintained by Long-Standing Balancing Selection and May Confer Susceptibility to Multiple Sclerosis. Molecular Biology and Evolution, 2012, 29, 1599-1613.	8.9	27
201	Brain temperature. NeuroReport, 2012, 23, 483-487.	1.2	27
202	iPSC-derived LewisX+CXCR4+Î²1-integrin+ neural stem cells improve the amyotrophic lateral sclerosis phenotype by preserving motor neurons and muscle innervation in human and rodent models. Human Molecular Genetics, 2016, 25, 3152-3163.	2.9	27
203	Glucose-6-Phosphate Dehydrogenase Lodi^844C: A Study on Its Expression in Blood Cells and Muscle. Enzyme, 1991, 45, 180-187.	0.7	26
204	A G+1->A transversion at the 5' splice site of intron 69 of the dystrophin gene causing the absence of peripheral nerve Dp 116 and severe clinical involvement in a DMD patient. Human Molecular Genetics, 1995, 4, 2171-2174.	2.9	26
205	Functional analysis of novel KCNQ2 and KCNQ3 gene variants found in a large pedigree with benign familial neonatal convulsions (BFNC). Neurogenetics, 2005, 6, 185-193.	1.4	26
206	The T-786C NOS3 polymorphism in Alzheimer's disease: Association and influence on gene expression. Neuroscience Letters, 2005, 382, 300-303.	2.1	26
207	ALS genetic modifiers that increase survival of SOD1 mice and are suitable for therapeutic development. Progress in Neurobiology, 2011, 95, 133-148.	5.7	26
208	The Brain is Hypothermic in Patients with Mitochondrial Diseases. Journal of Cerebral Blood Flow and Metabolism, 2014, 34, 915-920.	4.3	26
209	The influence of coenzyme Q10 on total serum calcium concentration in two patients with kearns-sayre syndrome and hypoparathyroidism. Neuromuscular Disorders, 1996, 6, 49-53.	0.6	25
210	Comparative Analysis of Vertebrate Dystrophin Loci Indicate Intron Gigantism as a Common Feature. Genome Research, 2003, 13, 764-772.	5.5	25
211	VCAM-1 expression on dystrophic muscle vessels has a critical role in the recruitment of human blood-derived CD133+ stem cells after intra-arterial transplantation. Blood, 2006, 108, 2857-66.	1.4	25
212	Increased brain temperature in Parkinsonâ€™s disease. NeuroReport, 2012, 23, 129-133.	1.2	25
213	Impairment of brain and muscle energy metabolism detected by magnetic resonance spectroscopy in hereditary spastic paraparesis type 28 patients with DDHD1 mutations. Journal of Neurology, 2014, 261, 1789-1793.	3.6	25
214	Mutational analysis of COQ2 in patients with MSA in Italy. Neurobiology of Aging, 2016, 45, 213.e1-213.e2.	3.1	25
215	Neurofascin (NFASC) gene mutation causes autosomal recessive ataxia with demyelinating neuropathy. Parkinsonism and Related Disorders, 2019, 63, 66-72.	2.2	25
216	Relevance of sequence and structure elements for deletion events in the dystrophin gene major hot-spot. Human Genetics, 2003, 112, 272-288.	3.8	24

#	Article	IF	Citations
217	Altered glutamate uptake in peripheral tissues from Down Syndrome patients. Neuroscience Letters, 2003, 343, 73-76.	2.1	24
218	Characterization of two novel SETX mutations in AOA2 patients reveals aspects of the pathophysiological role of senataxin. Neurogenetics, 2010, 11, 91-100.	1.4	24
219	Cerebrospinal fluid progranulin levels in patients with different multiple sclerosis subtypes. Neuroscience Letters, 2010, 469, 234-236.	2.1	24
220	Advances, Challenges, and Perspectives in Translational Stem Cell Therapy for Amyotrophic Lateral Sclerosis. Molecular Neurobiology, 2019, 56, 6703-6715.	4.0	24
221	Micro<scp>RNA</scp>s as regulators of cell death mechanisms in amyotrophic lateral sclerosis. Journal of Cellular and Molecular Medicine, 2019, 23, 1647-1656.	3.6	24
222	Comparative Analysis of the Human Dystrophin and Utrophin Gene Structures. Genetics, 2002, 160, 793-798.	2.9	24
223	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
224	Analysis of splicing parameters in the dystrophin gene: relevance for physiological and pathogenetic splicing mechanisms. Human Genetics, 2001, 109, 73-84.	3.8	23
225	New molecular findings in congenital myopathies due to selenoprotein N gene mutations. Journal of the Neurological Sciences, 2011, 300, 107-113.	0.6	23
226	Balancing selection is common in the extended MHC region but most alleles with opposite risk profile for autoimmune diseases are neutrally evolving. BMC Evolutionary Biology, 2011, 11, 171.	3.2	23
227	Specific profiles of neurocognitive and reading functions in a sample of 42 Italian boys with Duchenne Muscular Dystrophy. Child Neuropsychology, 2013, 19, 350-369.	1.3	23
228	Extended phenotype description and new molecular findings in late onset glycogen storage disease type II: a northern Italy population study and review of the literature. Journal of Neurology, 2014, 261, 83-97.	3.6	23
229	Chronic progressive external ophthalmoplegia: A correlative study of quantitative molecular data and histochemical and biochemical profile. Journal of the Neurological Sciences, 1994, 123, 140-146.	0.6	22
230	Cerebellar degeneration and hearing loss in a patient with idiopathic myenteric ganglionitis. European Journal of Gastroenterology and Hepatology, 2005, 17, 449-452.	1.6	22
231	Follow-up of a large population of asymptomatic/oligosymptomatic hyperckemic subjects. Journal of Neurology, 2006, 253, 1399-1403.	3.6	22
232	Pediatric Biobanking: A Pilot Qualitative Survey of Practices, Rules, and Researcher Opinions in Ten European Countries. Biopreservation and Biobanking, 2012, 10, 29-36.	1.0	22
233	Myotonia congenita: Novel mutations in CLCN1 gene and functional characterizations in Italian patients. Journal of the Neurological Sciences, 2012, 318, 65-71.	0.6	22
234	A novel homozygous PLA2G6 mutation causes dystonia-parkinsonism. Parkinsonism and Related Disorders, 2015, 21, 337-339.	2.2	22

#	Article	IF	Citations
235	Genome-wide RNA-seq of iPSC-derived motor neurons indicates selective cytoskeletal perturbation in Brownâ€“Vialetto disease that is partially rescued by riboflavin. Scientific Reports, 2017, 7, 46271.	3.3	22
236	A population genetics study of the Familial Mediterranean Fever gene: evidence of balancing selection under an overdominance regime. Genes and Immunity, 2009, 10, 678-686.	4.1	21
237	Ex vivo expansion of human circulating myogenic progenitors on cluster-assembled nanostructured TiO2. Biomaterials, 2010, 31, 5385-5396.	11.4	21
238	A Cortically Blind Patient With Preserved Visual Imagery. Cognitive and Behavioral Neurology, 2010, 23, 44-48.	0.9	21
239	Optic atrophy plus phenotype due to mutations in the OPA1 gene: Two more Italian families. Journal of the Neurological Sciences, 2012, 315, 146-149.	0.6	21
240	Evolutionary Analysis of the Contact System Indicates that Kininogen Evolved Adaptively in Mammals and in Human Populations. Molecular Biology and Evolution, 2013, 30, 1397-1408.	8.9	21
241	Lower motor neuron disease with respiratory failure caused by a novel MAPT mutation. Neurology, 2014, 82, 1990-1998.	1.1	21
242	Spinal muscular atrophy with respiratory distress type 1: Clinical phenotypes, molecular pathogenesis and therapeutic insights. Journal of Cellular and Molecular Medicine, 2020, 24, 1169-1178.	3.6	21
243	Transcriptional activation of the non-muscle, full-length dystrophin isoforms in Duchenne muscular dystrophy skeletal muscle. Journal of the Neurological Sciences, 2001, 186, 51-57.	0.6	20
244	GSK3Î² genetic variability in patients with Multiple Sclerosis. Neuroscience Letters, 2011, 497, 46-48.	2.1	20
245	Identification of a new susceptibility variant for multiple sclerosis in OAS1 by population genetics analysis. Human Genetics, 2012, 131, 87-97.	3.8	20
246	Direct Reprogramming of Adult Somatic Cells into other Lineages: Past Evidence and Future Perspectives. Cell Transplantation, 2013, 22, 921-944.	2.5	20
247	Molecular, genetic and stem cellâ€mediated therapeutic strategies for spinal muscular atrophy (<scp>SMA</scp>). Journal of Cellular and Molecular Medicine, 2014, 18, 187-196.	3.6	20
248	Stormorken Syndrome Caused by a p.R304W STIM1 Mutation: The First Italian Patient and a Review of the Literature. Frontiers in Neurology, 2018, 9, 859.	2.4	20
249	Disease Modeling and Therapeutic Strategies in CMT2A: State of the Art. Molecular Neurobiology, 2019, 56, 6460-6471.	4.0	20
250	A collection of 33 novel human mtDNA homoplasmic variants. Human Mutation, 2002, 20, 409-409.	2.5	19
251	Mitochondrial A12308G polymorphism affects clinical features in patients with single mtDNA macrodeletion. European Journal of Human Genetics, 2003, 11, 896-898.	2.8	19
252	An intragenic deletion/inversion event in the DMD gene determines a novel exon creation and results in a BMD phenotype. Human Genetics, 2004, 115, 13-18.	3.8	19

#	Article	IF	Citations
253	Clinical Studies in Stem Cells Transplantation for Stroke: A Review. Current Vascular Pharmacology, 2010, 8, 29-34.	1.7	19
254	Unusual adult-onset Leigh syndrome presentation due to the mitochondrial m.9176T>C mutation. Biochemical and Biophysical Research Communications, 2011, 412, 245-248.	2.1	19
255	Research advances in gene therapy approaches for the treatment of amyotrophic lateral sclerosis. Cellular and Molecular Life Sciences, 2012, 69, 1641-1650.	5.4	19
256	Multiple sclerosis and mitochondrial myopathy: An unusual combination of diseases. Journal of Neurology, 1994, 241, 511-516.	3.6	18
257	Chemokine network in multiple sclerosis: role in pathogenesis and targeting for future treatments. Expert Review of Neurotherapeutics, 2004, 4, 439-453.	2.8	18
258	Heparan sulfate proteoglycan induces the production of NO and TNF-alpha by murine microglia. Immunity and Ageing, 2005, 2, 11.	4.2	18
259	Absence of TREM2 polymorphisms in patients with Alzheimer's disease and Frontotemporal Lobar Degeneration. Neuroscience Letters, 2007, 411, 133-137.	2.1	18
260	No major progranulin genetic variability contribution to disease etiopathogenesis in an ALS Italian cohort. Neurobiology of Aging, 2011, 32, 1157-1158.	3.1	18
261	E-selectin A561C and G98T polymorphisms influence susceptibility and course of multiple sclerosis. Journal of Neuroimmunology, 2005, 165, 201-205.	2.3	17
262	Association of neuronal nitric oxide synthase C276T polymorphism with Alzheimerâ€™s disease. Journal of Neurology, 2005, 252, 985-986.	3.6	17
263	Candidate gene analysis of IP-10 gene in patients with Alzheimer's disease. Neuroscience Letters, 2006, 404, 217-221.	2.1	17
264	A region in the dystrophin gene major hot spot harbors a cluster of deletion breakpoints and generates doubleâ€strand breaks in yeast. FASEB Journal, 2006, 20, 1910-1912.	0.5	17
265	MCP-1 A-2518G Polymorphism: Effect on Susceptibility for Frontotemporal Lobar Degeneration and on Cerebrospinal Fluid MCP-1 Levels. Journal of Alzheimer's Disease, 2009, 17, 125-133.	2.6	17
266	Stem Cell Salvage of Injured Peripheral Nerve. Cell Transplantation, 2015, 24, 213-222.	2.5	17
267	New Mutations in NEB Gene Discovered by Targeted Next-Generation Sequencing in Nemaline Myopathy Italian Patients. Journal of Molecular Neuroscience, 2016, 59, 351-359.	2.3	17
268	Progressive Encephalomyelitis with Rigidity and Myoclonus Associated With Anti-GlyR Antibodies and Hodgkinâ€™s Lymphoma: A Case Report. Frontiers in Neurology, 2017, 8, 401.	2.4	17
269	Can Intestinal Pseudo-Obstruction Drive Recurrent Stroke-Like Episodes in Late-Onset MELAS Syndrome? A Case Report and Review of the Literature. Frontiers in Neurology, 2019, 10, 38.	2.4	17
270	Molecular Approaches for the Treatment of Pompe Disease. Molecular Neurobiology, 2020, 57, 1259-1280.	4.0	17

#	Article	IF	Citations
271	Posterior reversible encephalopathy syndrome and COVID-19: A series of 6 cases from Lombardy, Italy. ENeurologicalSci, 2021, 22, 100306.	1.3	17
272	Carnitine and acyltransferase in experimental neurogenic atrophies: changes with treatment. Journal of Neurology, 1984, 231, 170-175.	3.6	16
273	Trans -acting factors may cause dystrophin splicing misregulation in BMD skeletal muscles. FEBS Letters, 2003, 537, 30-34.	2.8	16
274	Developmental and tissue-specific regulation of a novel dysferlin isoform. Muscle and Nerve, 2004, 30, 366-374.	2.2	16
275	Ultrastructural Mitochondrial Abnormalities in Patients With Sporadic Amyotrophic Lateral Sclerosis. Archives of Neurology, 2011, 68, 1612.	4.5	16
276	Steroid-responsive Hashimoto encephalopathy mimicking Creutzfeldtâ€“Jakob disease. Neurological Sciences, 2011, 32, 719-722.	1.9	16
277	A 66-year-old patient with vanishing white matter disease due to the p.Ala87Val EIF2B3 mutation. Neurology, 2012, 79, 2077-2078.	1.1	16
278	Natural Selection at the Brush-Border: Adaptations to Carbohydrate Diets in Humans and Other Mammals. Genome Biology and Evolution, 2015, 7, 2569-2584.	2.5	16
279	SLC25A46 mutations in patients with Parkinson's Disease and optic atrophy. Parkinsonism and Related Disorders, 2020, 74, 1-5.	2.2	16
280	Alternative Sources of Neurons and Glia from Somatic Stem Cells. Cell Transplantation, 2002, 11, 25-34.	2.5	15
281	A case of CPT deficiency, homoplasmic mtDNA mutation and ragged red fibers at muscle biopsy. Journal of the Neurological Sciences, 2005, 239, 21-24.	0.6	15
282	Parkin polymorphisms and environmental exposure: Decrease in age at onset of Parkinson's disease. NeuroToxicology, 2007, 28, 698-701.	3.0	15
283	Muscle expression of glucoseâ€6â€phosphate dehydrogenase deficiency in different variants. Clinical Genetics, 1995, 48, 232-237.	2.0	15
284	Candidate gene analysis of semaphorins in patients with Alzheimerâ€™s disease. Neurological Sciences, 2010, 31, 169-173.	1.9	15
285	The mammalian complement system as an epitome of hostâ€“pathogen genetic conflicts. Molecular Ecology, 2016, 25, 1324-1339.	3.9	15
286	A de novo C19orf12 heterozygous mutation in a patient with MPAN. Parkinsonism and Related Disorders, 2018, 48, 109-111.	2.2	15
287	Mental health and coping strategies in families of children and young adults with muscular dystrophies. Journal of Neurology, 2020, 267, 2054-2069.	3.6	15
288	Incidence and Risk Factors of Motor Neuron Disease in the Venice and Padua Districts of Italy, 1972â€“1979. Neuroepidemiology, 1983, 2, 236-242.	2.3	14

#	Article	IF	Citations
289	Phosphoglycerate kinase deficiency myopathy: Biochemical and immunological studies of the mutant enzyme. Muscle and Nerve, 1984, 7, 542-551.	2.2	14
290	Multiple deletions of mitochondrial DNA in a patient with periodic attacks of paralysis. Journal of the Neurological Sciences, 1993, 117, 24-27.	0.6	14
291	Immunocytochemistry of muscle cytoskeletal proteins in acid maltase deficiency. Muscle and Nerve, 1994, 17, 655-661.	2.2	14
292	Progressive, isolated language disturbance: Its significance in a 65-year-old-man. A case report with implications for treatment and review of literature. Journal of the Neurological Sciences, 2006, 240, 45-51.	0.6	14
293	Optic Neuritis as Isolated Manifestation of Leptomeningeal Carcinomatosis: A Case Report and Systematic Review of Ocular Manifestations of Neoplastic Meningitis. Neurology Research International, 2013, 2013, 1-9.	1.3	14
294	Abnormal brain temperature in early-onset Parkinson's disease. Movement Disorders, 2016, 31, 425-426.	3.9	14
295	Novel donor splice site mutations of AGL gene in glycogen storage disease type IIIa. Journal of Inherited Metabolic Disease, 1999, 22, 762-763.	3.6	13
296	Over-representation of exonic splicing enhancers in human intronless genes suggests multiple functions in mRNA processing. Biochemical and Biophysical Research Communications, 2004, 322, 470-476.	2.1	13
297	P-selectin glycoprotein ligand-1 variable number of tandem repeats (VNTR) polymorphism in patients with multiple sclerosis. Neuroscience Letters, 2005, 388, 149-152.	2.1	13
298	SELPLG and SELP single-nucleotide polymorphisms in multiple sclerosis. Neuroscience Letters, 2006, 394, 92-96.	2.1	13
299	Polymorphisms in the CPB2 Gene Are Maintained by Balancing Selection and Result in Haplotype-Preferential Splicing of Exon 7. Molecular Biology and Evolution, 2010, 27, 1945-1954.	8.9	13
300	Noncoding RNAs in Duchenne and Becker muscular dystrophies: role in pathogenesis and future prognostic and therapeutic perspectives. Cellular and Molecular Life Sciences, 2020, 77, 4299-4313.	5.4	13
301	A Novel Homozygous <scp>VPS11</scp> Variant May Cause Generalized Dystonia. Annals of Neurology, 2021, 89, 834-839.	5.3	13
302	Clinical experience with topiramate dosing and serum levels in patients with epilepsy. Seizure: the Journal of the British Epilepsy Association, 2006, 15, 86-92.	2.0	12
303	Colocalization of ribonuclear inclusions with muscle blind like-proteins in a family with myotonic dystrophy type 2 associated with a short CCTG expansion. Journal of the Neurological Sciences, 2008, 275, 159-163.	0.6	12
304	Role of VEGF gene variability in longevity: A lesson from the Italian population. Neurobiology of Aging, 2008, 29, 1917-1922.	3.1	12
305	A complex selection signature at the human AVPR1B gene. BMC Evolutionary Biology, 2009, 9, 123.	3.2	12
306	VEGF Haplotypes are Associated with Increased Risk to Progressive Supranuclear Palsy and Corticobasal Syndrome. Journal of Alzheimer's Disease, 2010, 21, 87-94.	2.6	12

#	Article	IF	Citations
307	BAG1 is a Protective Factor for Sporadic Frontotemporal Lobar Degeneration but not for Alzheimer's Disease. Journal of Alzheimer's Disease, 2011, 23, 701-707.	2.6	12
308	A novel nonsense mutation in the APTX gene associated with delayed DNA singleâ€strand break removal fails to enhance sensitivity to different genotoxic agents. Human Mutation, 2011, 32, E2118-33.	2.5	12
309	Sciatic endometriosis presenting as periodic (catamenial) sciatic radiculopathy. Journal of Neurology, 2012, 259, 1470-1471.	3.6	12
310	A novel CCM1mutation associated with multiple cerebral and vertebral cavernous malformations. BMC Neurology, 2014, 14, 158.	1.8	12
311	Cell-penetrating peptide-conjugated Morpholino rescues SMA in a symptomatic preclinical model. Molecular Therapy, 2022, 30, 1288-1299.	8.2	12
312	Insights into the identification of a molecular signature for amyotrophic lateral sclerosis exploiting integrated microRNA profiling of iPSC-derived motor neurons and exosomes. Cellular and Molecular Life Sciences, 2022, 79, 189.	5.4	12
313	Phosphorylase isoenzymes of human brain. Neurochemical Pathology, 1983, 1, 171-178.	1.1	11
314	Rapid direct diagnosis of deletions carriers of Duchenne and Becker muscular dystrophies. Lancet, The, 1994, 344, 302-303.	13.7	11
315	Albuminoid Genes: Evolving at the Interface of Dispensability and Selection. Genome Biology and Evolution, 2014, 6, 2983-2997.	2.5	11
316	Novel Splice-Site Mutation in SMN1 Associated with a very Severe SMA-I Phenotype. Journal of Molecular Neuroscience, 2015, 56, 212-215.	2.3	11
317	Mutations in TMEM230 are rare in autosomal dominant Parkinson's disease. Parkinsonism and Related Disorders, 2017, 39, 87-88.	2.2	11
318	A new case of limb girdle muscular dystrophy 2G in a Greek patient, founder effect and review of the literature. Neuromuscular Disorders, 2018, 28, 532-537.	0.6	11
319	Current understanding of and emerging treatment options for spinal muscular atrophy with respiratory distress type 1 (SMARD1). Cellular and Molecular Life Sciences, 2020, 77, 3351-3367.	5.4	11
320	Post-Activation Brain Warming: A 1-H MRS Thermometry Study. PLoS ONE, 2015, 10, e0127314.	2.5	11
321	Alpha1-antichymotrypsin induces TNF-Î± production and NF-ÎºB activation in the murine N9 microglial cell line. Neuroscience Letters, 2009, 467, 40-42.	2.1	10
322	Variants in SNAP25 are targets of natural selection and influence verbal performances in women. Cellular and Molecular Life Sciences, 2012, 69, 1705-1715.	5.4	10
323	ISPD mutations account for a small proportion of Italian Limb Girdle Muscular Dystrophy cases. BMC Neurology, 2015, 15, 172.	1.8	10
324	Dystoniaâ€ataxia syndrome with permanent torsional nystagmus caused by ECHS1 deficiency. Annals of Clinical and Translational Neurology, 2020, 7, 839-845.	3.7	10

#	Article	IF	Citations
325	Clinical and genetic features of a cohort of patients with MFN2-related neuropathy. Scientific Reports, 2022, 12, 6181.	3.3	10
326	Cytochrome c oxidase during human fetal development. International Journal of Developmental Neuroscience, 1989, 7, 5-14.	1.6	9
327	Muscle glucose 6-phosphate dehydrogenase (G6PD) deficiency and oxidant stress during physical exercise. Cell Biochemistry and Function, 1995, 13, 297-298.	2.9	9
328	In Vitro and In Vivo Tetracycline-Controlled Myogenic Conversion of NIH-3T3 Cells: Evidence of Programmed Cell Death after Muscle Cell Transplantation. Cell Transplantation, 2001, 10, 209-221.	2.5	9
329	Correlation of Circulating CD133+ Progenitor Subclasses with a Mild Phenotype in Duchenne Muscular Dystrophy Patients. PLoS ONE, 2008, 3, e2218.	2.5	9
330	Is KIF24 a genetic risk factor for Frontotemporal Lobar Degeneration?. Neuroscience Letters, 2010, 482, 240-244.	2.1	9
331	Genetics and Expression Analysis of the Specificity Protein 4 Gene (SP4) in Patients with Alzheimer's Disease and Frontotemporal Lobar Degeneration. Journal of Alzheimer's Disease, 2012, 31, 537-542.	2.6	9
332	Central hyperthermia, brain hyperthermia and low hypothalamus temperature. Clinical Autonomic Research, 2012, 22, 299-301.	2.5	9
333	Diverse selective regimes shape genetic diversity atADARgenes and at their coding targets. RNA Biology, 2015, 12, 149-161.	3.1	9
334	Loss of the nucleoporin Aladin in central nervous system and fibroblasts of Allgrove Syndrome. Human Molecular Genetics, 2019, 28, 3921-3927.	2.9	9
335	Down's syndrome fibroblasts anticipate the accumulation of specific ageing-related mtDNA mutations. Annals of Neurology, 2001, 49, 137-138.	5.3	8
336	Mitochondrial-DNA nucleotides G4298A and T10010C as pathogenic mutations: the confirmation in two new cases. Mitochondrion, 2004, 3, 279-283.	3.4	8
337	Aphasic and visual aura with increased vasogenic leakage: An atypical migrainosus status. Journal of the Neurological Sciences, 2009, 285, 227-229.	0.6	8
338	In vitro analysis of splice site mutations in the CLCN1 gene using the minigene assay. Molecular Biology Reports, 2014, 41, 2865-2874.	2.3	8
339	Novel SETX variants in a patient with ataxia, neuropathy, and oculomotor apraxia are associated with normal sensitivity to oxidative DNA damaging agents. Brain and Development, 2014, 36, 682-689.	1.1	8
340	Paroxysmal Nocturnal Hemoglobinuria (Pnh): Brain Mri Ischemic Lesions In Neurologically Asymtomatic Patients. Scientific Reports, 2018, 8, 476.	3.3	8
341	Investigation of New Morpholino Oligomers to Increase Survival Motor Neuron Protein Levels in Spinal Muscular Atrophy. International Journal of Molecular Sciences, 2018, 19, 167.	4.1	8
342	CSF transplantation of a specific iPSC-derived neural stem cell subpopulation ameliorates the disease phenotype in a mouse model of spinal muscular atrophy with respiratory distress type 1. Experimental Neurology, 2019, 321, 113041.	4.1	8

#	Article	IF	Citations
343	Hereditary hemorrhagic telangiectasia associated with cortical development malformation due to a start loss mutation in ENG. BMC Neurology, 2020, 20, 316.	1.8	8
344	Unravelling Genetic Factors Underlying Corticobasal Syndrome: A Systematic Review. Cells, 2021, 10, 171.	4.1	8
345	Multiple neutral maltase activities in normal and acid maltase-deficient human muscle. Experimental Neurology, 1984, 84, 565-578.	4.1	7
346	Iron release and oxidant damage in human myoblasts by divicine. Life Sciences, 2000, 66, PL85-PL91.	4.3	7
347	Candidate gene analysis of selectin cluster in patients with multiple sclerosis. Journal of Neurology, 2009, 256, 832-833.	3.6	7
348	CCL8/MCP-2 association analysis in patients with Alzheimerâ€™s disease and frontotemporal lobar degeneration. Journal of Neurology, 2009, 256, 1379-1381.	3.6	7
349	Genetic variability in the ACE gene region surrounding the Alu I/D polymorphism is maintained by balancing selection in human populations. Pharmacogenetics and Genomics, 2010, 20, 131-134.	1.5	7
350	Two novel mutations in PEO1 (Twinkle) gene associated with chronic external ophthalmoplegia. Journal of the Neurological Sciences, 2011, 308, 173-176.	0.6	7
351	Long-Standing Balancing Selection in theTHBS4Gene: Influence on Sex-Specific Brain Expression and Gray Matter Volumes in Alzheimer Disease. Human Mutation, 2013, 34, 743-753.	2.5	7
352	Growing Evidence about the Relationship between Vessel Dissection and Scuba Diving. Case Reports in Neurology, 2013, 5, 155-161.	0.7	7
353	Impact of <scp>COVIDâ€19</scp> on the quality of life of patients with neuromuscular disorders in the <scp>L</scp>ombardy area, <scp>I</scp>taly. Muscle and Nerve, 2021, 64, 474-482.	2.2	7
354	Congenital muscular dystrophy with muscle inflammation alpha dystroglycan glycosylation defect and no mutation in FKRP gene. Journal of the Neurological Sciences, 2006, 243, 47-51.	0.6	6
355	Language Disturbances in a Group of Participants Suffering from Duchenne Muscular Dystrophy: A Pilot Study. Perceptual and Motor Skills, 2007, 104, 663-676.	1.3	6
356	Candidate gene analysis of SPARCL1 gene in patients with multiple sclerosis. Neuroscience Letters, 2007, 425, 173-176.	2.1	6
357	FUS/TLS Genetic Variability in Sporadic Frontotemporal Lobar Degeneration. Journal of Alzheimer's Disease, 2010, 19, 1317-1322.	2.6	6
358	Acute rhabdomyolysis induced by tonicâ€“clonic epileptic seizures in a patient with glucose-6-phosphate dehydrogenase deficiency. Journal of Neurology, 2013, 260, 2669-2671.	3.6	6
359	Central Nervous System Involvement in Common Variable Immunodeficiency: A Case of Acute Unilateral Optic Neuritis in a 26-Year-Old Italian Patient. Frontiers in Neurology, 2018, 9, 1031.	2.4	6
360	Bilateral Cavernous Carotid Aneurysms: Atypical Presentation of a Rare Cause of Mass Effect. A Case Report and a Review of the Literature. Frontiers in Neurology, 2018, 9, 619.	2.4	6

#	Article	IF	Citations
361	Animal Models of CMT2A: State-of-art and Therapeutic Implications. Molecular Neurobiology, 2020, 57, 5121-5129.	4.0	6
362	Clinical and biochemical evidence of skeletal muscle involvement in galactose-1-phosphate uridyl transferase deficiency. Journal of Neurology, 1993, 240, 272-277.	3.6	5
363	Migraine, stroke and patent foramen ovale: a dangerous trio?. Journal of Cardiovascular Medicine, 2008, 9, 233-238.	1.5	5
364	Charcot-Marie-Tooth type 1a in a child with Long QT syndrome. European Journal of Paediatric Neurology, 2009, 13, 459-462.	1.6	5
365	Transthyretin asn90 variant: Amyloidogenic or non-amyloidogenic role. Journal of the Neurological Sciences, 2009, 284, 113-115.	0.6	5
366	An evolutionary history of the selectin gene cluster in humans. Heredity, 2012, 109, 117-126.	2.6	5
367	Insights into disease mechanisms and potential therapeutics for C9orf72-related amyotrophic lateral sclerosis/frontotemporal dementia. Ageing Research Reviews, 2020, 64, 101172.	10.9	5
368	Clinical, neuroradiological and genetic findings in a cohort of patients with multiple Cerebral Cavernous Malformations. Metabolic Brain Disease, 2021, 36, 1871-1878.	2.9	5
369	Ryanodine receptor gene point mutation and malignant hyperthermia susceptibility. Journal of Neurology, 1995, 242, 127-133.	3.6	4
370	The novel mitochondrial tRNAAsn gene mutation m.5709T>C produces ophthalmoparesis and respiratory impairment. European Journal of Human Genetics, 2012, 20, 357-360.	2.8	4
371	Glucose-free/high-protein diet improves hepatomegaly and exercise intolerance in glycogen storage disease type III mice. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 3407-3417.	3.8	4
372	Ophthalmoplegia Due to Miller Fisher Syndrome in a Patient With Myasthenia Gravis. Frontiers in Neurology, 2019, 10, 823.	2.4	4
373	Case Report: Efficacy of Rituximab in a Patient With Familial Mediterranean Fever and Multiple Sclerosis. Frontiers in Neurology, 2020, 11, 591395.	2.4	4
374	Charcotâ€“Marieâ€“Tooth disease type 2F associated with biallelic HSPB1 mutations. Annals of Clinical and Translational Neurology, 2021, 8, 1158-1164.	3.7	4
375	Usefulness of Multi-Parametric MRI for the Investigation of Posterior Cortical Atrophy. PLoS ONE, 2015, 10, e0140639.	2.5	4
376	Intra-aortic injection of myoblasts in mdx mice: Genetic and technetium-99m cell labeling and biodistribution. , 1997, 20, 757-759.		3
377	Two dystrophin proteins and transcripts in a mild dystrophinopathic patient. Neuromuscular Disorders, 2003, 13, 13-16.	0.6	3
378	Is erythropoietin gene a modifier factor in amyotrophic lateral sclerosis?. Neurobiology of Aging, 2009, 30, 842-844.	3.1	3

#	Article	IF	Citations
379	Beta-lactam antibiotic offers neuroprotection in a spinal muscular atrophy model by multiple mechanisms. Annals of Neurosciences, 2011, 18, 156-7.	1.7	3
380	Brain temperature in multiple sclerosis. Multiple Sclerosis Journal, 2014, 20, 894-896.	3.0	3
381	Reply: DGUOK recessive mutations in patients with CPEO, mitochondrial myopathy, parkinsonism and mtDNA deletions. Brain, 2018, 141, e4-e4.	7.6	3
382	Subclinical Leberâ€™s hereditary optic neuropathy with pediatric acute spinal cord onset: more than meets the eye. BMC Neurology, 2018, 18, 220.	1.8	3
383	Hyperacute extensive spinal cord infarction and negative spine magnetic resonance imaging: a case report and review of the literature. Medicine (United States), 2020, 99, e22900.	1.0	3
384	A case report of late-onset cerebellar ataxia associated with a rare p.R342W TGM6 (SCA35) mutation. BMC Neurology, 2020, 20, 408.	1.8	3
385	Microscopic Polyangiitis With Selective Involvement of Central and Peripheral Nervous System: A Case Report. Frontiers in Neurology, 2020, 11, 269.	2.4	3
386	Early Findings in Neonatal Cases of RYR1â€“Related Congenital Myopathies. Frontiers in Neurology, 2021, 12, 664618.	2.4	3
387	Molecular analysis of SMARD1 patient-derived cells demonstrates that nonsense-mediated mRNA decay is impaired. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, 908-910.	1.9	3
388	Sporadic Distal Myopathy with Early Adult Onset: Study of Muscle Biopsies and Muscle Cell Cultures. European Neurology, 1989, 29, 287-290.	1.4	2
389	Muscular Dystrophy: Central Nervous System Î±-Dystroglycan Glycosylation Defects and Brain Malformation. Journal of Child Neurology, 2010, 25, 312-320.	1.4	2
390	No association of IFI16 (interferon-inducible protein 16) variants with susceptibility to multiple sclerosis. Journal of Neuroimmunology, 2014, 271, 49-52.	2.3	2
391	Anti-sulfatide reactivity in patients with celiac disease. Scandinavian Journal of Gastroenterology, 2017, 52, 409-413.	1.5	2
392	Clinical Reasoning: A 75-year-old man with parkinsonism, mood depression, and weight loss. Neurology, 2018, 90, 572-575.	1.1	2
393	Herpes Simplex virus type 2 myeloradiculitis with a pure motor presentation in a liver transplant recipient. Transplant Infectious Disease, 2020, 22, e13236.	1.7	2
394	Safety and efficacy of rt-PA treatment for acute stroke in pseudoxanthoma elasticum: the first report. Journal of Thrombosis and Thrombolysis, 2021, 51, 176-179.	2.1	2
395	Sodium Levels Predict Disability at Discharge in Guillain-BarrÃ© Syndrome: A Retrospective Cohort Study. Frontiers in Neurology, 2021, 12, 729252.	2.4	2
396	CACNA1S mutation associated with a case of juvenile-onset congenital myopathy. Journal of the Neurological Sciences, 2021, 431, 120047.	0.6	2

#	Article	IF	Citations
397	Limb girdle muscular dystrophy due to gene mutations: new mutations expand the clinical spectrum of a still challenging diagnosis. Acta Myologica, 2020, 39, 67-82.	1.5	2
398	Muscle glucose-6-phosphate dehydrogenase deficiency: Restoration of enzymatic activity in hybrid myotubes. Muscle and Nerve, 1993, 16, 594-600.	2.2	1
399	Insertion of Two Independent Enhancers in the Long Terminal Repeat of a Self-Inactivating Vector Results in High-Titer Retroviral Vectors with Tissue-Specific Expression. Human Gene Therapy, 1998, 9, 2459-2468.	2.7	1
400	Cytochrome c oxidase deficiency. International Review of Neurobiology, 2002, 53, 205-240.	2.0	1
401	The m.12316G>A mutation in the mitochondrial tRNALeu(CUN) gene is associated with mitochondrial myopathy and respiratory impairment. Journal of the Neurological Sciences, 2010, 292, 107-110.	0.6	1
402	Cell-dependent kinase inhibitor 2A and 2B genetic variability in patients with Alzheimerâ€™s disease. Journal of Neurology, 2011, 258, 704-705.	3.6	1
403	Expanding the clinical spectrum of the mitochondrial mutation A13084T in the ND5 gene. Neurology: Genetics, 2020, 6, e511.	1.9	1
404	In vivo biolistic technique in control and mdx dystrophic mice. , 1996, 19, 912-914.		0
405	The role of interleukin-6 (IL-6) in the proliferation and differentiation of human neural stem cells. Neuroscience Research Communications, 2001, 29, 1-14.	0.2	0
406	Safety of Systemic Chemotherapy in a Patient With Mitochondrial Myopathy and Nonâ€“Small-Cell Lung Cancer. Journal of Clinical Oncology, 2012, 30, e226-e228.	1.6	0
407	Late-onset leukoencephalopathy in a patient with recessive EARS2 mutations. Neurology: Genetics, 2020, 6, e488.	1.9	0
408	Expanding the genotypic and phenotypic spectrum of Betaâ€propeller poteinâ€associated neurodegeneration. European Journal of Neurology, 2021, 28, e25-e27.	3.3	0