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	Cell-penetrating peptide-conjugated Morpholino rescues SMA in a symptomatic preclinical model. Molecular Therapy, 2022, 30, 1288-1299.	8.2	12
2	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
3	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
4	Clinical and genetic features of a cohort of patients with MFN2-related neuropathy. Scientific Reports, 2022, 12, 6181.	3.3	10
5	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
6	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
7	Expanding the genotypic and phenotypic spectrum of Betaâ€propeller poteinâ€associated neurodegeneration. European Journal of Neurology, 2021, 28, e25-e27.	3.3	0
8	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
9	Unravelling Genetic Factors Underlying Corticobasal Syndrome: A Systematic Review. Cells, 2021, 10, 171.	4.1	8
10	A Novel Homozygous <scp>VPS11</scp> Variant May Cause Generalized Dystonia. Annals of Neurology, 2021, 89, 834-839.	5.3	13
11	Posterior reversible encephalopathy syndrome and COVID-19: A series of 6 cases from Lombardy, Italy. ENeurologicalSci, 2021, 22, 100306.	1.3	17
12	Charcotâ€“Marieâ€“Tooth disease type 2F associated with biallelic HSPB1 mutations. Annals of Clinical and Translational Neurology, 2021, 8, 1158-1164.	3.7	4
13	Early Findings in Neonatal Cases of RYR1â€“Related Congenital Myopathies. Frontiers in Neurology, 2021, 12, 664618.	2.4	3
14	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
15	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
16	Sodium Levels Predict Disability at Discharge in Guillain-BarrÃ© Syndrome: A Retrospective Cohort Study. Frontiers in Neurology, 2021, 12, 729252.	2.4	2
17	CACNA1S mutation associated with a case of juvenile-onset congenital myopathy. Journal of the Neurological Sciences, 2021, 431, 120047.	0.6	2
18	Molecular Approaches for the Treatment of Pompe Disease. Molecular Neurobiology, 2020, 57, 1259-1280.	4.0	17

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19	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
20	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
21	Expanding the clinical spectrum of the mitochondrial mutation A13084T in the ND5 gene. Neurology: Genetics, 2020, 6, e511.	1.9	1
22	Dystoniaâ€ataxia syndrome with permanent torsional nystagmus caused by ECHS1 deficiency. Annals of Clinical and Translational Neurology, 2020, 7, 839-845.	3.7	10
23	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
24	Insights into disease mechanisms and potential therapeutics for C9orf72-related amyotrophic lateral sclerosis/frontotemporal dementia. Ageing Research Reviews, 2020, 64, 101172.	10.9	5
25	Animal Models of CMT2A: State-of-art and Therapeutic Implications. Molecular Neurobiology, 2020, 57, 5121-5129.	4.0	6
26	Hereditary hemorrhagic telangiectasia associated with cortical development malformation due to a start loss mutation in ENG. BMC Neurology, 2020, 20, 316.	1.8	8
27	Late-onset leukoencephalopathy in a patient with recessive EARS2 mutations. Neurology: Genetics, 2020, 6, e488.	1.9	0
28	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
29	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
30	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
31	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
32	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
33	Microscopic Polyangiitis With Selective Involvement of Central and Peripheral Nervous System: A Case Report. Frontiers in Neurology, 2020, 11, 269.	2.4	3
34	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
35	Neural Stem Cell Transplantation for Neurodegenerative Diseases. International Journal of Molecular Sciences, 2020, 21, 3103.	4.1	105
36	Glial cells involvement in spinal muscular atrophy: Could SMA be a neuroinflammatory disease?. Neurobiology of Disease, 2020, 140, 104870.	4.4	35

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37	SLC25A46 mutations in patients with Parkinson's Disease and optic atrophy. Parkinsonism and Related Disorders, 2020, 74, 1-5.	2.2	16
38	Case Report: Efficacy of Rituximab in a Patient With Familial Mediterranean Fever and Multiple Sclerosis. Frontiers in Neurology, 2020, 11, 591395.	2.4	4
39	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
40	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
41	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
42	R-Loops in Motor Neuron Diseases. Molecular Neurobiology, 2019, 56, 2579-2589.	4.0	39
43	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
44	Loss of the nucleoporin Aladin in central nervous system and fibroblasts of Allgrove Syndrome. Human Molecular Genetics, 2019, 28, 3921-3927.	2.9	9
45	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
46	Ophthalmoplegia Due to Miller Fisher Syndrome in a Patient With Myasthenia Gravis. Frontiers in Neurology, 2019, 10, 823.	2.4	4
47	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
48	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
49	Disease Modeling and Therapeutic Strategies in CMT2A: State of the Art. Molecular Neurobiology, 2019, 56, 6460-6471.	4.0	20
50	Neurofascin (NFASC) gene mutation causes autosomal recessive ataxia with demyelinating neuropathy. Parkinsonism and Related Disorders, 2019, 63, 66-72.	2.2	25
51	Advances, Challenges, and Perspectives in Translational Stem Cell Therapy for Amyotrophic Lateral Sclerosis. Molecular Neurobiology, 2019, 56, 6703-6715.	4.0	24
52	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
53	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
54	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

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55	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
56	Reply: DGUOK recessive mutations in patients with CPEO, mitochondrial myopathy, parkinsonism and mtDNA deletions. Brain, 2018, 141, e4-e4.	7.6	3
57	A de novo C19orf12 heterozygous mutation in a patient with MPAN. Parkinsonism and Related Disorders, 2018, 48, 109-111.	2.2	15
58	Paroxysmal Nocturnal Hemoglobinuria (Pnh): Brain Mri Ischemic Lesions In Neurologically Asymtomatic Patients. Scientific Reports, 2018, 8, 476.	3.3	8
59	Clinical Reasoning: A 75-year-old man with parkinsonism, mood depression, and weight loss. Neurology, 2018, 90, 572-575.	1.1	2
60	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
61	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
62	Mitochondrial dysfunction in fibroblasts of Multiple System Atrophy. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 3588-3597.	3.8	32
63	Mitochondrial Dysregulation and Impaired Autophagy in iPSC-Derived Dopaminergic Neurons of Multiple System Atrophy. Stem Cell Reports, 2018, 11, 1185-1198.	4.8	46
64	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
65	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
66	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
67	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
68	Brain Mitochondria, Aging, and Parkinsonâ€™s Disease. Genes, 2018, 9, 250.	2.4	53
69	MicroRNA expression analysis identifies a subset of downregulated miRNAs in ALS motor neuron progenitors. Scientific Reports, 2018, 8, 10105.	3.3	53
70	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
71	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
72	The italian limb girdle muscular dystrophy registry: Relative frequency, clinical features, and differential diagnosis. Muscle and Nerve, 2017, 55, 55-68.	2.2	86

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73	Anti-sulfatide reactivity in patients with celiac disease. Scandinavian Journal of Gastroenterology, 2017, 52, 409-413.	1.5	2
74	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
75	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
76	Mutations in TMEM230 are rare in autosomal dominant Parkinson's disease. Parkinsonism and Related Disorders, 2017, 39, 87-88.	2.2	11
77	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
78	Development of Therapeutics for C9ORF72 ALS/FTD-Related Disorders. Molecular Neurobiology, 2017, 54, 4466-4476.	4.0	30
79	Clinical and Paraclinical Indicators of Motor System Impairment in Hereditary Spastic Paraplegia: A Pilot Study. PLoS ONE, 2016, 11, e0153283.	2.5	41
80	The mammalian complement system as an epitome of hostâ€“pathogen genetic conflicts. Molecular Ecology, 2016, 25, 1324-1339.	3.9	15
81	Abnormal brain temperature in early-onset Parkinson's disease. Movement Disorders, 2016, 31, 425-426.	3.9	14
82	The evolutionary history of genes involved in spoken and written language: beyond FOXP2. Scientific Reports, 2016, 6, 22157.	3.3	55
83	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
84	Mutational analysis of COQ2 in patients with MSA in Italy. Neurobiology of Aging, 2016, 45, 213.e1-213.e2.	3.1	25
85	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
86	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
87	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
88	ISPD mutations account for a small proportion of Italian Limb Girdle Muscular Dystrophy cases. BMC Neurology, 2015, 15, 172.	1.8	10
89	Stem Cell Salvage of Injured Peripheral Nerve. Cell Transplantation, 2015, 24, 213-222.	2.5	17
90	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

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91	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
92	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
93	MFN2-related neuropathies: Clinical features, molecular pathogenesis and therapeutic perspectives. Journal of the Neurological Sciences, 2015, 356, 7-18.	0.6	112
94	OASes and STING: Adaptive Evolution in Concert. Genome Biology and Evolution, 2015, 7, 1016-1032.	2.5	57
95	A novel homozygous PLA2G6 mutation causes dystonia-parkinsonism. Parkinsonism and Related Disorders, 2015, 21, 337-339.	2.2	22
96	Diverse selective regimes shape genetic diversity atADARgenes and at their coding targets. RNA Biology, 2015, 12, 149-161.	3.1	9
97	Variants in KIF1A gene in dominant and sporadic forms of hereditary spastic paraparesis. Journal of Neurology, 2015, 262, 2684-2690.	3.6	55
98	CHCHD10mutations in Italian patients with sporadic amyotrophic lateral sclerosis: Figure 1. Brain, 2015, 138, e372-e372.	7.6	59
99	Post-Activation Brain Warming: A 1-H MRS Thermometry Study. PLoS ONE, 2015, 10, e0127314.	2.5	11
100	Usefulness of Multi-Parametric MRI for the Investigation of Posterior Cortical Atrophy. PLoS ONE, 2015, 10, e0140639.	2.5	4
101	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
102	An Evolutionary Analysis of Antigen Processing and Presentation across Different Timescales Reveals Pervasive Selection. PLoS Genetics, 2014, 10, e1004189.	3.5	42
103	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
104	Albuminoid Genes: Evolving at the Interface of Dispensability and Selection. Genome Biology and Evolution, 2014, 6, 2983-2997.	2.5	11
105	Lower motor neuron disease with respiratory failure caused by a novel MAPT mutation. Neurology, 2014, 82, 1990-1998.	1.1	21
106	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
107	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
108	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

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109	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
110	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
111	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
112	Antisense Oligonucleotide Therapy for the Treatment of C9ORF72 ALS/FTD Diseases. Molecular Neurobiology, 2014, 50, 721-732.	4.0	48
113	Molecular Therapeutic Strategies for Spinal Muscular Atrophies: Current and Future Clinical Trials. Clinical Therapeutics, 2014, 36, 128-140.	2.5	74
114	Induced neural stem cells: Methods of reprogramming and potential therapeutic applications. Progress in Neurobiology, 2014, 114, 15-24.	5.7	39
115	Brain temperature in multiple sclerosis. Multiple Sclerosis Journal, 2014, 20, 894-896.	3.0	3
116	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
117	Subclinical leukodystrophy and infertility in a man with a novel homozygous CLCN2 mutation. Neurology, 2014, 83, 1217-1218.	1.1	42
118	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
119	A novel CCM1mutation associated with multiple cerebral and vertebral cavernous malformations. BMC Neurology, 2014, 14, 158.	1.8	12
120	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
121	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
122	The Brain is Hypothermic in Patients with Mitochondrial Diseases. Journal of Cerebral Blood Flow and Metabolism, 2014, 34, 915-920.	4.3	26
123	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
124	No association of IFI16 (interferon-inducible protein 16) variants with susceptibility to multiple sclerosis. Journal of Neuroimmunology, 2014, 271, 49-52.	2.3	2
125	Ongoing therapeutic trials and outcome measures for Duchenne muscular dystrophy. Cellular and Molecular Life Sciences, 2013, 70, 4585-4602.	5.4	53
126	Crohn's Disease Loci Are Common Targets of Protozoa-Driven Selection. Molecular Biology and Evolution, 2013, 30, 1077-1087.	8.9	28

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127	Progranulin gene (GRN) promoter methylation is increased in patients with sporadic frontotemporal lobar degeneration. Neurological Sciences, 2013, 34, 899-903.	1.9	30
128	Direct Reprogramming of Adult Somatic Cells into other Lineages: Past Evidence and Future Perspectives. Cell Transplantation, 2013, 22, 921-944.	2.5	20
129	Mutations in DNA2 Link Progressive Myopathy to Mitochondrial DNA Instability. American Journal of Human Genetics, 2013, 92, 293-300.	6.2	115
130	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
131	Decreased circulating miRNA levels in patients with primary progressive multiple sclerosis. Multiple Sclerosis Journal, 2013, 19, 1938-1942.	3.0	98
132	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
133	Defective autophagy in spastizin mutated patients with hereditary spastic paraparesis type 15. Brain, 2013, 136, 3119-3139.	7.6	74
134	Mitochondrial Fusion Proteins and Human Diseases. Neurology Research International, 2013, 2013, 1-11.	1.3	85
135	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
136	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
137	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
138	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
139	Growing Evidence about the Relationship between Vessel Dissection and Scuba Diving. Case Reports in Neurology, 2013, 5, 155-161.	0.7	7
140	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
141	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
142	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
143	Incontinence in Late-Onset Pompe Disease: An Underdiagnosed Treatable Condition. European Neurology, 2012, 68, 75-78.	1.4	27
144	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

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145	Next-generation sequencing reveals DGUOK mutations in adult patients with mitochondrial DNA multiple deletions. Brain, 2012, 135, 3404-3415.	7.6	81
146	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
147	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
148	Increased brain temperature in Parkinsonâ€™s disease. NeuroReport, 2012, 23, 129-133.	1.2	25
149	Brain temperature. NeuroReport, 2012, 23, 483-487.	1.2	27
150	Genetic Correction of Human Induced Pluripotent Stem Cells from Patients with Spinal Muscular Atrophy. Science Translational Medicine, 2012, 4, 165ra162.	12.4	180
151	Central hyperthermia, brain hyperthermia and low hypothalamus temperature. Clinical Autonomic Research, 2012, 22, 299-301.	2.5	9
152	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
153	Quantitative muscle strength assessment in duchenne muscular dystrophy: longitudinal study and correlation with functional measures. BMC Neurology, 2012, 12, 91.	1.8	52
154	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
155	Selective DNA Methylation of BDNF Promoter in Bipolar Disorder: Differences Among Patients with BDI and BDII. Neuropsychopharmacology, 2012, 37, 1647-1655.	5.4	166
156	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
157	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
158	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
159	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
160	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
161	Progranulin Gene Variability and Plasma Levels in Bipolar Disorder and Schizophrenia. PLoS ONE, 2012, 7, e32164.	2.5	34
162	An evolutionary history of the selectin gene cluster in humans. Heredity, 2012, 109, 117-126.	2.6	5

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163	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
164	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
165	Sciatic endometriosis presenting as periodic (catamenial) sciatic radiculopathy. Journal of Neurology, 2012, 259, 1470-1471.	3.6	12
166	Direct reprogramming of human astrocytes into neural stem cells and neurons. Experimental Cell Research, 2012, 318, 1528-1541.	2.6	143
167	Identification of a new susceptibility variant for multiple sclerosis in OAS1 by population genetics analysis. Human Genetics, 2012, 131, 87-97.	3.8	20
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