

# Nereo Bresolin

## List of Publications by Year in descending order

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Version: 2024-02-01

408  
papers

18,295  
citations

11608

70  
h-index

24915

109  
g-index

412  
all docs

412  
docs citations

412  
times ranked

24819  
citing authors

#	ARTICLE	IF	CITATIONS
1	Cell-penetrating peptide-conjugated Morpholino rescues SMA in a symptomatic preclinical model. <i>Molecular Therapy</i> , 2022, 30, 1288-1299.	3.7	12
2	Molecular analysis of SMARD1 patient-derived cells demonstrates that nonsense-mediated mRNA decay is impaired. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2022, 93, 908-910.	0.9	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. <i>Cellular and Molecular Life Sciences</i> , 2022, 79, 189.	2.4	12
4	Clinical and genetic features of a cohort of patients with MFN2-related neuropathy. <i>Scientific Reports</i> , 2022, 12, 6181.	1.6	10
5	Safety and efficacy of rt-PA treatment for acute stroke in pseudoxanthoma elasticum: the first report. <i>Journal of Thrombosis and Thrombolysis</i> , 2021, 51, 176-179.	1.0	2
6	Extracellular vesicles and amyotrophic lateral sclerosis: from misfolded protein vehicles to promising clinical biomarkers. <i>Cellular and Molecular Life Sciences</i> , 2021, 78, 561-572.	2.4	42
7	Expanding the genotypic and phenotypic spectrum of Beta-protein-associated neurodegeneration. <i>European Journal of Neurology</i> , 2021, 28, e25-e27.	1.7	0
8	Management of patients with neuromuscular disorders at the time of the SARS-CoV-2 pandemic. <i>Journal of Neurology</i> , 2021, 268, 1580-1591.	1.8	34
9	Unravelling Genetic Factors Underlying Corticobasal Syndrome: A Systematic Review. <i>Cells</i> , 2021, 10, 171.	1.8	8
10	A Novel Homozygous <i>VPS11</i> Variant May Cause Generalized Dystonia. <i>Annals of Neurology</i> , 2021, 89, 834-839.	2.8	13
11	Posterior reversible encephalopathy syndrome and COVID-19: A series of 6 cases from Lombardy, Italy. <i>ENeurologicalSci</i> , 2021, 22, 100306.	0.5	17
12	Charcot-Marie-Tooth disease type 2F associated with biallelic <i>HSPB1</i> mutations. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 1158-1164.	1.7	4
13	Early Findings in Neonatal Cases of RYR1-Related Congenital Myopathies. <i>Frontiers in Neurology</i> , 2021, 12, 664618.	1.1	3
14	Impact of COVID-19 on the quality of life of patients with neuromuscular disorders in the Lombardy area, Italy. <i>Muscle and Nerve</i> , 2021, 64, 474-482.	1.0	7
15	Clinical, neuroradiological and genetic findings in a cohort of patients with multiple Cerebral Cavernous Malformations. <i>Metabolic Brain Disease</i> , 2021, 36, 1871-1878.	1.4	5
16	Sodium Levels Predict Disability at Discharge in Guillain-Barré Syndrome: A Retrospective Cohort Study. <i>Frontiers in Neurology</i> , 2021, 12, 729252.	1.1	2
17	CACNA1S mutation associated with a case of juvenile-onset congenital myopathy. <i>Journal of the Neurological Sciences</i> , 2021, 431, 120047.	0.3	2
18	Molecular Approaches for the Treatment of Pompe Disease. <i>Molecular Neurobiology</i> , 2020, 57, 1259-1280.	1.9	17

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19	Spinal muscular atrophy with respiratory distress type 1: Clinical phenotypes, molecular pathogenesis and therapeutic insights. <i>Journal of Cellular and Molecular Medicine</i> , 2020, 24, 1169-1178.	1.6	21
20	Herpes Simplex virus type 2 myeloradiculitis with a pure motor presentation in a liver transplant recipient. <i>Transplant Infectious Disease</i> , 2020, 22, e13236.	0.7	2
21	Expanding the clinical spectrum of the mitochondrial mutation A13084T in the <i>ND5</i> gene. <i>Neurology: Genetics</i> , 2020, 6, e511.	0.9	1
22	Dystonia-ataxia syndrome with permanent torsional nystagmus caused by ECHS1 deficiency. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 839-845.	1.7	10
23	Hyperacute extensive spinal cord infarction and negative spine magnetic resonance imaging: a case report and review of the literature. <i>Medicine (United States)</i> , 2020, 99, e22900.	0.4	3
24	Insights into disease mechanisms and potential therapeutics for C9orf72-related amyotrophic lateral sclerosis/frontotemporal dementia. <i>Ageing Research Reviews</i> , 2020, 64, 101172.	5.0	5
25	Animal Models of CMT2A: State-of-art and Therapeutic Implications. <i>Molecular Neurobiology</i> , 2020, 57, 5121-5129.	1.9	6
26	Hereditary hemorrhagic telangiectasia associated with cortical development malformation due to a start loss mutation in ENG. <i>BMC Neurology</i> , 2020, 20, 316.	0.8	8
27	Late-onset leukoencephalopathy in a patient with recessive EARS2 mutations. <i>Neurology: Genetics</i> , 2020, 6, e488.	0.9	0
28	A case report of late-onset cerebellar ataxia associated with a rare p.R342W TGM6 (SCA35) mutation. <i>BMC Neurology</i> , 2020, 20, 408.	0.8	3
29	Mental health and coping strategies in families of children and young adults with muscular dystrophies. <i>Journal of Neurology</i> , 2020, 267, 2054-2069.	1.8	15
30	Silence superoxide dismutase 1 (SOD1): a promising therapeutic target for amyotrophic lateral sclerosis (ALS). <i>Expert Opinion on Therapeutic Targets</i> , 2020, 24, 295-310.	1.5	49
31	Nusinersen treatment and cerebrospinal fluid neurofilaments: An explorative study on Spinal Muscular Atrophy type 3 patients. <i>Journal of Cellular and Molecular Medicine</i> , 2020, 24, 3034-3039.	1.6	47
32	Current understanding of and emerging treatment options for spinal muscular atrophy with respiratory distress type 1 (SMARD1). <i>Cellular and Molecular Life Sciences</i> , 2020, 77, 3351-3367.	2.4	11
33	Microscopic Polyangiitis With Selective Involvement of Central and Peripheral Nervous System: A Case Report. <i>Frontiers in Neurology</i> , 2020, 11, 269.	1.1	3
34	Noncoding RNAs in Duchenne and Becker muscular dystrophies: role in pathogenesis and future prognostic and therapeutic perspectives. <i>Cellular and Molecular Life Sciences</i> , 2020, 77, 4299-4313.	2.4	13
35	Neural Stem Cell Transplantation for Neurodegenerative Diseases. <i>International Journal of Molecular Sciences</i> , 2020, 21, 3103.	1.8	105
36	Glial cells involvement in spinal muscular atrophy: Could SMA be a neuroinflammatory disease?. <i>Neurobiology of Disease</i> , 2020, 140, 104870.	2.1	35

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37	SLC25A46 mutations in patients with Parkinson's Disease and optic atrophy. <i>Parkinsonism and Related Disorders</i> , 2020, 74, 1-5.	1.1	16
38	Case Report: Efficacy of Rituximab in a Patient With Familial Mediterranean Fever and Multiple Sclerosis. <i>Frontiers in Neurology</i> , 2020, 11, 591395.	1.1	4
39	The Role of Mitochondria in Neurodegenerative Diseases: the Lesson from Alzheimer's Disease and Parkinson's Disease. <i>Molecular Neurobiology</i> , 2020, 57, 2959-2980.	1.9	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. <i>Acta Myologica</i> , 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. <i>Acta Myologica</i> , 2020, 39, 67-82.	1.5	2
42	R-Loops in Motor Neuron Diseases. <i>Molecular Neurobiology</i> , 2019, 56, 2579-2589.	1.9	39
43	Preconditioning and Cellular Engineering to Increase the Survival of Transplanted Neural Stem Cells for Motor Neuron Disease Therapy. <i>Molecular Neurobiology</i> , 2019, 56, 3356-3367.	1.9	36
44	Loss of the nucleoporin Aladin in central nervous system and fibroblasts of Allgrove Syndrome. <i>Human Molecular Genetics</i> , 2019, 28, 3921-3927.	1.4	9
45	Diagnostic and Prognostic Role of Blood and Cerebrospinal Fluid and Blood Neurofilaments in Amyotrophic Lateral Sclerosis: A Review of the Literature. <i>International Journal of Molecular Sciences</i> , 2019, 20, 4152.	1.8	47
46	Ophthalmoplegia Due to Miller Fisher Syndrome in a Patient With Myasthenia Gravis. <i>Frontiers in Neurology</i> , 2019, 10, 823.	1.1	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. <i>Experimental Neurology</i> , 2019, 321, 113041.	2.0	8
48	Human induced pluripotent stem cell models for the study and treatment of Duchenne and Becker muscular dystrophies. <i>Therapeutic Advances in Neurological Disorders</i> , 2019, 12, 175628641983347.	1.5	32
49	Disease Modeling and Therapeutic Strategies in CMT2A: State of the Art. <i>Molecular Neurobiology</i> , 2019, 56, 6460-6471.	1.9	20
50	Neurofascin (NFASC) gene mutation causes autosomal recessive ataxia with demyelinating neuropathy. <i>Parkinsonism and Related Disorders</i> , 2019, 63, 66-72.	1.1	25
51	Advances, Challenges, and Perspectives in Translational Stem Cell Therapy for Amyotrophic Lateral Sclerosis. <i>Molecular Neurobiology</i> , 2019, 56, 6703-6715.	1.9	24
52	Can Intestinal Pseudo-Obstruction Drive Recurrent Stroke-Like Episodes in Late-Onset MELAS Syndrome? A Case Report and Review of the Literature. <i>Frontiers in Neurology</i> , 2019, 10, 38.	1.1	17
53	“Ears of the Lynx” MRI Sign Is Associated with SPG11 and SPG15 Hereditary Spastic Paraplegia. <i>American Journal of Neuroradiology</i> , 2019, 40, 199-203.	1.2	50
54	MicroRNAs as regulators of cell death mechanisms in amyotrophic lateral sclerosis. <i>Journal of Cellular and Molecular Medicine</i> , 2019, 23, 1647-1656.	1.6	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. <i>Brain</i> , 2019, 142, 276-294.	3.7	31
56	Reply: DGUOK recessive mutations in patients with CPEO, mitochondrial myopathy, parkinsonism and mtDNA deletions. <i>Brain</i> , 2018, 141, e4-e4.	3.7	3
57	A de novo C19orf12 heterozygous mutation in a patient with MPAN. <i>Parkinsonism and Related Disorders</i> , 2018, 48, 109-111.	1.1	15
58	Paroxysmal Nocturnal Hemoglobinuria (Pnh): Brain Mri Ischemic Lesions In Neurologically Asymptomatic Patients. <i>Scientific Reports</i> , 2018, 8, 476.	1.6	8
59	Clinical Reasoning: A 75-year-old man with parkinsonism, mood depression, and weight loss. <i>Neurology</i> , 2018, 90, 572-575.	1.5	2
60	Subclinical Leber's hereditary optic neuropathy with pediatric acute spinal cord onset: more than meets the eye. <i>BMC Neurology</i> , 2018, 18, 220.	0.8	3
61	Central Nervous System Involvement in Common Variable Immunodeficiency: A Case of Acute Unilateral Optic Neuritis in a 26-Year-Old Italian Patient. <i>Frontiers in Neurology</i> , 2018, 9, 1031.	1.1	6
62	Mitochondrial dysfunction in fibroblasts of Multiple System Atrophy. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2018, 1864, 3588-3597.	1.8	32
63	Mitochondrial Dysregulation and Impaired Autophagy in iPSC-Derived Dopaminergic Neurons of Multiple System Atrophy. <i>Stem Cell Reports</i> , 2018, 11, 1185-1198.	2.3	46
64	Stormorken Syndrome Caused by a p.R304W STIM1 Mutation: The First Italian Patient and a Review of the Literature. <i>Frontiers in Neurology</i> , 2018, 9, 859.	1.1	20
65	Bilateral Cavernous Carotid Aneurysms: Atypical Presentation of a Rare Cause of Mass Effect. A Case Report and a Review of the Literature. <i>Frontiers in Neurology</i> , 2018, 9, 619.	1.1	6
66	A new case of limb girdle muscular dystrophy 2G in a Greek patient, founder effect and review of the literature. <i>Neuromuscular Disorders</i> , 2018, 28, 532-537.	0.3	11
67	Glucose-free/high-protein diet improves hepatomegaly and exercise intolerance in glycogen storage disease type III mice. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2018, 1864, 3407-3417.	1.8	4
68	Brain Mitochondria, Aging, and Parkinson's Disease. <i>Genes</i> , 2018, 9, 250.	1.0	53
69	MicroRNA expression analysis identifies a subset of downregulated miRNAs in ALS motor neuron progenitors. <i>Scientific Reports</i> , 2018, 8, 10105.	1.6	53
70	Investigation of New Morpholino Oligomers to Increase Survival Motor Neuron Protein Levels in Spinal Muscular Atrophy. <i>International Journal of Molecular Sciences</i> , 2018, 19, 167.	1.8	8
71	Multiparametric quantitative MRI assessment of thigh muscles in limb-girdle muscular dystrophy 2A and 2B. <i>Muscle and Nerve</i> , 2018, 58, 550-558.	1.0	37
72	The Italian limb girdle muscular dystrophy registry: Relative frequency, clinical features, and differential diagnosis. <i>Muscle and Nerve</i> , 2017, 55, 55-68.	1.0	86

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73	Anti-sulfatide reactivity in patients with celiac disease. <i>Scandinavian Journal of Gastroenterology</i> , 2017, 52, 409-413.	0.6	2
74	Genome-wide RNA-seq of iPSC-derived motor neurons indicates selective cytoskeletal perturbation in Brownâ€™Violetto disease that is partially rescued by riboflavin. <i>Scientific Reports</i> , 2017, 7, 46271.	1.6	22
75	Assessing mental health in boys with Duchenne muscular dystrophy: Emotional, behavioural and neurodevelopmental profile in an Italian clinical sample. <i>European Journal of Paediatric Neurology</i> , 2017, 21, 639-647.	0.7	46
76	Mutations in TMEM230 are rare in autosomal dominant Parkinson's disease. <i>Parkinsonism and Related Disorders</i> , 2017, 39, 87-88.	1.1	11
77	Progressive Encephalomyelitis with Rigidity and Myoclonus Associated With Anti-GlyR Antibodies and Hodgkinâ€™s Lymphoma: A Case Report. <i>Frontiers in Neurology</i> , 2017, 8, 401.	1.1	17
78	Development of Therapeutics for C9ORF72 ALS/FTD-Related Disorders. <i>Molecular Neurobiology</i> , 2017, 54, 4466-4476.	1.9	30
79	Clinical and Paraclinical Indicators of Motor System Impairment in Hereditary Spastic Paraplegia: A Pilot Study. <i>PLoS ONE</i> , 2016, 11, e0153283.	1.1	41
80	The mammalian complement system as an epitome of hostâ€™pathogen genetic conflicts. <i>Molecular Ecology</i> , 2016, 25, 1324-1339.	2.0	15
81	Abnormal brain temperature in early-onset Parkinson's disease. <i>Movement Disorders</i> , 2016, 31, 425-426.	2.2	14
82	The evolutionary history of genes involved in spoken and written language: beyond FOXP2. <i>Scientific Reports</i> , 2016, 6, 22157.	1.6	55
83	New Mutations in NEB Gene Discovered by Targeted Next-Generation Sequencing in Nemaline Myopathy Italian Patients. <i>Journal of Molecular Neuroscience</i> , 2016, 59, 351-359.	1.1	17
84	Mutational analysis of COQ2 in patients with MSA in Italy. <i>Neurobiology of Aging</i> , 2016, 45, 213.e1-213.e2.	1.5	25
85	Selective mitochondrial depletion, apoptosis resistance, and increased mitophagy in human Charcot-Marie-Tooth 2A motor neurons. <i>Human Molecular Genetics</i> , 2016, 25, 4266-4281.	1.4	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. <i>Human Molecular Genetics</i> , 2016, 25, 3152-3163.	1.4	27
87	Spinal muscular atrophy phenotype is ameliorated in human motor neurons by SMN increase via different novel RNA therapeutic approaches. <i>Scientific Reports</i> , 2015, 5, 11746.	1.6	37
88	ISPD mutations account for a small proportion of Italian Limb Girdle Muscular Dystrophy cases. <i>BMC Neurology</i> , 2015, 15, 172.	0.8	10
89	Stem Cell Salvage of Injured Peripheral Nerve. <i>Cell Transplantation</i> , 2015, 24, 213-222.	1.2	17
90	Gene therapy rescues disease phenotype in a spinal muscular atrophy with respiratory distress type 1 (SMARD1) mouse model. <i>Science Advances</i> , 2015, 1, e1500078.	4.7	33

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91	Natural Selection at the Brush-Border: Adaptations to Carbohydrate Diets in Humans and Other Mammals. <i>Genome Biology and Evolution</i> , 2015, 7, 2569-2584.	1.1	16
92	Novel Splice-Site Mutation in SMN1 Associated with a very Severe SMA-I Phenotype. <i>Journal of Molecular Neuroscience</i> , 2015, 56, 212-215.	1.1	11
93	MFN2-related neuropathies: Clinical features, molecular pathogenesis and therapeutic perspectives. <i>Journal of the Neurological Sciences</i> , 2015, 356, 7-18.	0.3	112
94	OASes and STING: Adaptive Evolution in Concert. <i>Genome Biology and Evolution</i> , 2015, 7, 1016-1032.	1.1	57
95	A novel homozygous PLA2G6 mutation causes dystonia-parkinsonism. <i>Parkinsonism and Related Disorders</i> , 2015, 21, 337-339.	1.1	22
96	Diverse selective regimes shape genetic diversity at <i>ADAR</i> genes and at their coding targets. <i>RNA Biology</i> , 2015, 12, 149-161.	1.5	9
97	Variants in KIF1A gene in dominant and sporadic forms of hereditary spastic paraparesis. <i>Journal of Neurology</i> , 2015, 262, 2684-2690.	1.8	55
98	CHCHD10 mutations in Italian patients with sporadic amyotrophic lateral sclerosis: Figure 1. <i>Brain</i> , 2015, 138, e372-e372.	3.7	59
99	Post-Activation Brain Warming: A 1-H MRS Thermometry Study. <i>PLoS ONE</i> , 2015, 10, e0127314.	1.1	11
100	Usefulness of Multi-Parametric MRI for the Investigation of Posterior Cortical Atrophy. <i>PLoS ONE</i> , 2015, 10, e0140639.	1.1	4
101	Genetic adaptation of the human circadian clock to day-length latitudinal variations and relevance for affective disorders. <i>Genome Biology</i> , 2014, 15, 499.	3.8	28
102	An Evolutionary Analysis of Antigen Processing and Presentation across Different Timescales Reveals Pervasive Selection. <i>PLoS Genetics</i> , 2014, 10, e1004189.	1.5	42
103	Ancient and Recent Selective Pressures Shaped Genetic Diversity at AIM2-Like Nucleic Acid Sensors. <i>Genome Biology and Evolution</i> , 2014, 6, 830-845.	1.1	28
104	Albuminoid Genes: Evolving at the Interface of Dispensability and Selection. <i>Genome Biology and Evolution</i> , 2014, 6, 2983-2997.	1.1	11
105	Lower motor neuron disease with respiratory failure caused by a novel <i>MAPT</i> mutation. <i>Neurology</i> , 2014, 82, 1990-1998.	1.5	21
106	Minimally invasive transplantation of iPSC-derived ALDHhiSSCloVLA4+ neural stem cells effectively improves the phenotype of an amyotrophic lateral sclerosis model. <i>Human Molecular Genetics</i> , 2014, 23, 342-354.	1.4	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. <i>Journal of Neurology</i> , 2014, 261, 83-97.	1.8	23
108	Stem cell transplantation for amyotrophic lateral sclerosis: therapeutic potential and perspectives on clinical translation. <i>Cellular and Molecular Life Sciences</i> , 2014, 71, 3257-3268.	2.4	32

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109	Mutations in CYP2U1, DDHD2 and GBA2 genes are rare causes of complicated forms of hereditary spastic paraparesis. <i>Journal of Neurology</i> , 2014, 261, 373-381.	1.8	62
110	Effect of Combined Systemic and Local Morpholino Treatment on the Spinal Muscular Atrophy $\hat{7}$ Mouse Model Phenotype. <i>Clinical Therapeutics</i> , 2014, 36, 340-356.e5.	1.1	44
111	Molecular, genetic and stem cell-mediated therapeutic strategies for spinal muscular atrophy (<scp>SMA</scp>). <i>Journal of Cellular and Molecular Medicine</i> , 2014, 18, 187-196.	1.6	20
112	Antisense Oligonucleotide Therapy for the Treatment of C9ORF72 ALS/FTD Diseases. <i>Molecular Neurobiology</i> , 2014, 50, 721-732.	1.9	48
113	Molecular Therapeutic Strategies for Spinal Muscular Atrophies: Current and Future Clinical Trials. <i>Clinical Therapeutics</i> , 2014, 36, 128-140.	1.1	74
114	Induced neural stem cells: Methods of reprogramming and potential therapeutic applications. <i>Progress in Neurobiology</i> , 2014, 114, 15-24.	2.8	39
115	Brain temperature in multiple sclerosis. <i>Multiple Sclerosis Journal</i> , 2014, 20, 894-896.	1.4	3
116	Glycogen storage disease type III: A novel Agl knockout mouse model. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2014, 1842, 2318-2328.	1.8	28
117	Subclinical leukodystrophy and infertility in a man with a novel homozygous <i>CLCN2</i> mutation. <i>Neurology</i> , 2014, 83, 1217-1218.	1.5	42
118	iPSC-Derived Neural Stem Cells Act via Kinase Inhibition to Exert Neuroprotective Effects in Spinal Muscular Atrophy with Respiratory Distress Type 1. <i>Stem Cell Reports</i> , 2014, 3, 297-311.	2.3	34
119	A novel CCM1 mutation associated with multiple cerebral and vertebral cavernous malformations. <i>BMC Neurology</i> , 2014, 14, 158.	0.8	12
120	Impairment of brain and muscle energy metabolism detected by magnetic resonance spectroscopy in hereditary spastic paraparesis type 28 patients with DDHD1 mutations. <i>Journal of Neurology</i> , 2014, 261, 1789-1793.	1.8	25
121	In vitro analysis of splice site mutations in the CLCN1 gene using the minigene assay. <i>Molecular Biology Reports</i> , 2014, 41, 2865-2874.	1.0	8
122	The Brain is Hypothermic in Patients with Mitochondrial Diseases. <i>Journal of Cerebral Blood Flow and Metabolism</i> , 2014, 34, 915-920.	2.4	26
123	Novel SETX variants in a patient with ataxia, neuropathy, and oculomotor apraxia are associated with normal sensitivity to oxidative DNA damaging agents. <i>Brain and Development</i> , 2014, 36, 682-689.	0.6	8
124	No association of IFI16 (interferon-inducible protein 16) variants with susceptibility to multiple sclerosis. <i>Journal of Neuroimmunology</i> , 2014, 271, 49-52.	1.1	2
125	Ongoing therapeutic trials and outcome measures for Duchenne muscular dystrophy. <i>Cellular and Molecular Life Sciences</i> , 2013, 70, 4585-4602.	2.4	53
126	Crohn's Disease Loci Are Common Targets of Protozoa-Driven Selection. <i>Molecular Biology and Evolution</i> , 2013, 30, 1077-1087.	3.5	28



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127	Progranulin gene (GRN) promoter methylation is increased in patients with sporadic frontotemporal lobar degeneration. <i>Neurological Sciences</i> , 2013, 34, 899-903.	0.9	30
128	Direct Reprogramming of Adult Somatic Cells into other Lineages: Past Evidence and Future Perspectives. <i>Cell Transplantation</i> , 2013, 22, 921-944.	1.2	20
129	Mutations in DNA2 Link Progressive Myopathy to Mitochondrial DNA Instability. <i>American Journal of Human Genetics</i> , 2013, 92, 293-300.	2.6	115
130	A 175 Million Year History of T Cell Regulatory Molecules Reveals Widespread Selection, with Adaptive Evolution of Disease Alleles. <i>Immunity</i> , 2013, 38, 1129-1141.	6.6	30
131	Decreased circulating miRNA levels in patients with primary progressive multiple sclerosis. <i>Multiple Sclerosis Journal</i> , 2013, 19, 1938-1942.	1.4	98
132	Acute rhabdomyolysis induced by tonic-clonic epileptic seizures in a patient with glucose-6-phosphate dehydrogenase deficiency. <i>Journal of Neurology</i> , 2013, 260, 2669-2671.	1.8	6
133	Defective autophagy in spastizin mutated patients with hereditary spastic paraparesis type 15. <i>Brain</i> , 2013, 136, 3119-3139.	3.7	74
134	Mitochondrial Fusion Proteins and Human Diseases. <i>Neurology Research International</i> , 2013, 2013, 1-11.	0.5	85
135	Optic Neuritis as Isolated Manifestation of Leptomeningeal Carcinomatosis: A Case Report and Systematic Review of Ocular Manifestations of Neoplastic Meningitis. <i>Neurology Research International</i> , 2013, 2013, 1-9.	0.5	14
136	Long-Standing Balancing Selection in the THBS4 Gene: Influence on Sex-Specific Brain Expression and Gray Matter Volumes in Alzheimer Disease. <i>Human Mutation</i> , 2013, 34, 743-753.	1.1	7
137	Evolutionary Analysis of the Contact System Indicates that Kininogen Evolved Adaptively in Mammals and in Human Populations. <i>Molecular Biology and Evolution</i> , 2013, 30, 1397-1408.	3.5	21
138	Specific profiles of neurocognitive and reading functions in a sample of 42 Italian boys with Duchenne Muscular Dystrophy. <i>Child Neuropsychology</i> , 2013, 19, 350-369.	0.8	23
139	Growing Evidence about the Relationship between Vessel Dissection and Scuba Diving. <i>Case Reports in Neurology</i> , 2013, 5, 155-161.	0.3	7
140	Safety of Systemic Chemotherapy in a Patient With Mitochondrial Myopathy and Non-Small-Cell Lung Cancer. <i>Journal of Clinical Oncology</i> , 2012, 30, e226-e228.	0.8	0
141	The novel mitochondrial tRNA <sup>Asn</sup> gene mutation m.5709T>C produces ophthalmoparesis and respiratory impairment. <i>European Journal of Human Genetics</i> , 2012, 20, 357-360.	1.4	4
142	Pediatric Biobanking: A Pilot Qualitative Survey of Practices, Rules, and Researcher Opinions in Ten European Countries. <i>Biopreservation and Biobanking</i> , 2012, 10, 29-36.	0.5	22
143	Incontinence in Late-Onset Pompe Disease: An Underdiagnosed Treatable Condition. <i>European Neurology</i> , 2012, 68, 75-78.	0.6	27
144	A 66-year-old patient with vanishing white matter disease due to the p.Ala87Val EIF2B3 mutation. <i>Neurology</i> , 2012, 79, 2077-2078.	1.5	16

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145	Next-generation sequencing reveals DGUOK mutations in adult patients with mitochondrial DNA multiple deletions. <i>Brain</i> , 2012, 135, 3404-3415.	3.7	81
146	A Trans-Specific Polymorphism in ZC3HAV1 Is Maintained by Long-Standing Balancing Selection and May Confer Susceptibility to Multiple Sclerosis. <i>Molecular Biology and Evolution</i> , 2012, 29, 1599-1613.	3.5	27
147	Genetics and Expression Analysis of the Specificity Protein 4 Gene (SP4) in Patients with Alzheimer's Disease and Frontotemporal Lobar Degeneration. <i>Journal of Alzheimer's Disease</i> , 2012, 31, 537-542.	1.2	9
148	Increased brain temperature in Parkinson's disease. <i>NeuroReport</i> , 2012, 23, 129-133.	0.6	25
149	Brain temperature. <i>NeuroReport</i> , 2012, 23, 483-487.	0.6	27
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