Stefania Corti

List of Publications by Year
in descending order

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233 papers	9,309 citations	31902 53 h-index	60497 81 g-index
235 all docs	235 docs citations	235 times ranked	13452 citing authors

#	Article	IF	Citations
1	Cell-penetrating peptide-conjugated Morpholino rescues SMA in a symptomatic preclinical model. Molecular Therapy, 2022, 30, 1288-1299.	3.7	12
2	Mitochondrial DNA homeostasis impairment and dopaminergic dysfunction: A trembling balance. Ageing Research Reviews, 2022, 76, 101578.	5.0	15
3	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.	0.9	3
4	Targeting PTB for Glia-to-Neuron Reprogramming In Vitro and In Vivo for Therapeutic Development in Neurological Diseases. Biomedicines, 2022, 10, 399.	1.4	6
5	Case Report: Thymidine Kinase 2 (TK2) Deficiency: A Novel Mutation Associated With Childhood-Onset Mitochondrial Myopathy and Atypical Progression. Frontiers in Neurology, 2022, 13, 857279.	1.1	0
6	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.	2.4	12
7	Stathmins and Motor Neuron Diseases: Pathophysiology and Therapeutic Targets. Biomedicines, 2022, 10, 711.	1.4	9
8	Generation of two hiPSC lines (UMILi027-A and UMILi028-A) from early and late-onset Congenital Central hypoventilation Syndrome (CCHS) patients carrying a polyalanine expansion mutation in the PHOX2B gene. Stem Cell Research, 2022, 61, 102781.	0.3	0
9	Homozygous SOD1 Variation L144S Produces a Severe Form of Amyotrophic Lateral Sclerosis in an Iranian Family. Neurology: Genetics, 2022, 8, e645.	0.9	6
10	Clinical and genetic features of a cohort of patients with MFN2-related neuropathy. Scientific Reports, 2022, 12, 6181.	1.6	10
11	Inhibition of myostatin and related signaling pathways for the treatment of muscle atrophy in motor neuron diseases. Cellular and Molecular Life Sciences, 2022, 79, .	2.4	16
12	New Insights into Cerebral Vessel Disease Landscapes at Single-Cell Resolution: Pathogenetic and Therapeutic Perspectives. Biomedicines, 2022, 10, 1693.	1.4	1
13	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.	1.0	2
14	Extracellular vesicles and amyotrophic lateral sclerosis: from misfolded protein vehicles to promising clinical biomarkers. Cellular and Molecular Life Sciences, 2021, 78, 561-572.	2.4	42
15	Expanding the genotypic and phenotypic spectrum of Betaâ€propeller poteinâ€associated neurodegeneration. European Journal of Neurology, 2021, 28, e25-e27.	1.7	0
16	Management of patients with neuromuscular disorders at the time of the SARS-CoV-2 pandemic. Journal of Neurology, 2021, 268, 1580-1591.	1.8	34
17	A Novel Homozygous <scp>VPS11</scp> Variant May Cause Generalized Dystonia. Annals of Neurology, 2021, 89, 834-839.	2.8	13
18	Diagnostic and prognostic value of CSF neurofilaments in a cohort of patients with motor neuron disease: A crossâ€sectional study. Journal of Cellular and Molecular Medicine, 2021, 25, 3765-3771.	1.6	10

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19	Advancing Drug Discovery for Neurological Disorders Using iPSC-Derived Neural Organoids. International Journal of Molecular Sciences, 2021, 22, 2659.	1.8	33
20	Impairment of the neurotrophic signaling hub B-Raf contributes to motoneuron degeneration in spinal muscular atrophy. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, e2007785118.	3.3	11
21	Dysregulation of Muscle-Specific MicroRNAs as Common Pathogenic Feature Associated with Muscle Atrophy in ALS, SMA and SBMA: Evidence from Animal Models and Human Patients. International Journal of Molecular Sciences, 2021, 22, 5673.	1.8	14
22	Charcotâ€“Marieâ€“Tooth disease type 2F associated with biallelic HSPB1 mutations. Annals of Clinical and Translational Neurology, 2021, 8, 1158-1164.	1.7	4
23	Early Findings in Neonatal Cases of RYR1â€“Related Congenital Myopathies. Frontiers in Neurology, 2021, 12, 664618.	1.1	3
24	Sumoylation regulates the assembly and activity of the SMN complex. Nature Communications, 2021, 12, 5040.	5.8	8
25	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.	1.0	7
26	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. JAMA Neurology, 2021, 78, 1236.	4.5	46
27	Screening of LRP10 mutations in Parkinson's disease patients from Italy. Parkinsonism and Related Disorders, 2021, 89, 17-21.	1.1	5
28	Perspectives on hiPSC-Derived Muscle Cells as Drug Discovery Models for Muscular Dystrophies. International Journal of Molecular Sciences, 2021, 22, 9630.	1.8	3
29	Sodium Levels Predict Disability at Discharge in Guillain-BarrÃ© Syndrome: A Retrospective Cohort Study. Frontiers in Neurology, 2021, 12, 729252.	1.1	2
30	Onasemnogene abeparvovec gene therapy for symptomatic infantile-onset spinal muscular atrophy type 1 (STR1VE-EU): an open-label, single-arm, multicentre, phase 3 trial. Lancet Neurology, The, 2021, 20, 832-841.	4.9	112
31	CACNA1S mutation associated with a case of juvenile-onset congenital myopathy. Journal of the Neurological Sciences, 2021, 431, 120047.	0.3	2
32	Adeno-Associated Virus (AAV)-Mediated Gene Therapy for Duchenne Muscular Dystrophy: The Issue of Transgene Persistence. Frontiers in Neurology, 2021, 12, 814174.	1.1	27
33	Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. Nature Genetics, 2021, 53, 1636-1648.	9.4	223
34	Molecular Approaches for the Treatment of Pompe Disease. Molecular Neurobiology, 2020, 57, 1259-1280.	1.9	17
35	Back to the origins: Human brain organoids to investigate neurodegeneration. Brain Research, 2020, 1727, 146561.	1.1	12
36	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.	1.6	21

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37	STIM1 R304W in mice causes subgingival hair growth and an increased fraction of trabecular bone. Cell Calcium, 2020, 85, 102110.	1.1	8
38	Herpes Simplex virus type 2 myeloradiculitis with a pure motor presentation in a liver transplant recipient. Transplant Infectious Disease, 2020, 22, e13236.	0.7	2
39	Expanding the clinical spectrum of the mitochondrial mutation A13084T in the ND5 gene. Neurology: Genetics, 2020, 6, e511.	0.9	1
40	Dystoniaâ€ataxia syndrome with permanent torsional nystagmus caused by ECHS1 deficiency. Annals of Clinical and Translational Neurology, 2020, 7, 839-845.	1.7	10
41	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.	0.4	3
42	Insights into disease mechanisms and potential therapeutics for C9orf72-related amyotrophic lateral sclerosis/frontotemporal dementia. Ageing Research Reviews, 2020, 64, 101172.	5.0	5
43	Animal Models of CMT2A: State-of-art and Therapeutic Implications. Molecular Neurobiology, 2020, 57, 5121-5129.	1.9	6
44	Spinal muscular atrophy â€" challenges in the therapeutic era. Nature Reviews Neurology, 2020, 16, 655-656.	4.9	1
45	Systematic elucidation of neuron-astrocyte interaction in models of amyotrophic lateral sclerosis using multi-modal integrated bioinformatics workflow. Nature Communications, 2020, 11, 5579.	5.8	28
46	A case report of late-onset cerebellar ataxia associated with a rare p.R342W TGM6 (SCA35) mutation. BMC Neurology, 2020, 20, 408.	0.8	3
47	Silence superoxide dismutase 1 (SOD1): a promising therapeutic target for amyotrophic lateral sclerosis (ALS). Expert Opinion on Therapeutic Targets, 2020, 24, 295-310.	1.5	49
48	Nusinersen treatment and cerebrospinal fluid neurofilaments: An explorative study on Spinal Muscular Atrophy type 3 patients. Journal of Cellular and Molecular Medicine, 2020, 24, 3034-3039.	1.6	47
49	Synaptotagmin 13 is neuroprotective across motor neuron diseases. Acta Neuropathologica, 2020, 139, 837-853.	3.9	28
50	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.	2.4	11
51	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.	2.4	13
52	Neural Stem Cell Transplantation for Neurodegenerative Diseases. International Journal of Molecular Sciences, 2020, 21, 3103.	1.8	105
53	miR-129-5p: A key factor and therapeutic target in amyotrophic lateral sclerosis. Progress in Neurobiology, 2020, 190, 101803.	2.8	31
54	Glial cells involvement in spinal muscular atrophy: Could SMA be a neuroinflammatory disease?. Neurobiology of Disease, 2020, 140, 104870.	2.1	35

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55	SLC25A46 mutations in patients with Parkinson's Disease and optic atrophy. Parkinsonism and Related Disorders, 2020, 74, 1-5.	1.1	16
56	R-Loops in Motor Neuron Diseases. Molecular Neurobiology, 2019, 56, 2579-2589.	1.9	39
57	Preconditioning and Cellular Engineering to Increase the Survival of Transplanted Neural Stem Cells for Motor Neuron Disease Therapy. Molecular Neurobiology, 2019, 56, 3356-3367.	1.9	36
58	Loss of the nucleoporin Aladin in central nervous system and fibroblasts of Allgrove Syndrome. Human Molecular Genetics, 2019, 28, 3921-3927.	1.4	9
59	Diagnostic and Prognostic Role of Blood and Cerebrospinal Fluid and Blood Neurofilaments in Amyotrophic Lateral Sclerosis: A Review of the Literature. International Journal of Molecular Sciences, 2019, 20, 4152.	1.8	47
60	Ophthalmoplegia Due to Miller Fisher Syndrome in a Patient With Myasthenia Gravis. Frontiers in Neurology, 2019, 10, 823.	1.1	4
61	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.	2.0	8
62	Human induced pluripotent stem cell models for the study and treatment of Duchenne and Becker muscular dystrophies. Therapeutic Advances in Neurological Disorders, 2019, 12, 175628641983347.	1.5	32
63	Disease Modeling and Therapeutic Strategies in CMT2A: State of the Art. Molecular Neurobiology, 2019, 56, 6460-6471.	1.9	20
64	Neurofascin (NFASC) gene mutation causes autosomal recessive ataxia with demyelinating neuropathy. Parkinsonism and Related Disorders, 2019, 63, 66-72.	1.1	25
65	Advances, Challenges, and Perspectives in Translational Stem Cell Therapy for Amyotrophic Lateral Sclerosis. Molecular Neurobiology, 2019, 56, 6703-6715.	1.9	24
66	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.	1.1	17
67	iPSCs-Based Neural 3D Systems: A Multidimensional Approach for Disease Modeling and Drug Discovery. Cells, 2019, 8, 1438.	1.8	41
68	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.	1.6	24
69	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.	3.7	31
70	Study Design of STR1VE-EU, a Phase 3 Trial of AVXS-101 Gene-Replacement Therapy (GRT) in Patients With Spinal Muscular Atrophy Type 1 (SMA1) in Europe. , 2019, 50, .		0
71	mi<scp>RNA</scp> in spinal muscular atrophy pathogenesis and therapy. Journal of Cellular and Molecular Medicine, 2018, 22, 755-767.	1.6	46
72	In vitro models of multiple system atrophy from primary cells to induced pluripotent stem cells. Journal of Cellular and Molecular Medicine, 2018, 22, 2536-2546.	1.6	11

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73	Pregnancy outcomes in women with spinal muscular atrophy: A review. Journal of the Neurological Sciences, 2018, 388, 50-60.	0.3	14
74	Advances in spinal muscular atrophy therapeutics. Therapeutic Advances in Neurological Disorders, 2018, 11, 175628561875450.	1.5	69
75	Reply: DGUOK recessive mutations in patients with CPEO, mitochondrial myopathy, parkinsonism and mtDNA deletions. Brain, 2018, 141, e4-e4.	3.7	3
76	In Vivo Transient and Partial Cell Reprogramming to Pluripotency as a Therapeutic Tool for Neurodegenerative Diseases. Molecular Neurobiology, 2018, 55, 6850-6862.	1.9	12
77	A de novo C19orf12 heterozygous mutation in a patient with MPAN. Parkinsonism and Related Disorders, 2018, 48, 109-111.	1.1	15
78	Time Is Motor Neuron: Therapeutic Window and Its Correlation with Pathogenetic Mechanisms in Spinal Muscular Atrophy. Molecular Neurobiology, 2018, 55, 6307-6318.	1.9	53
79	Therapeutic Strategies Under Development Targeting Inflammatory Mechanisms in Amyotrophic Lateral Sclerosis. Molecular Neurobiology, 2018, 55, 2789-2813.	1.9	32
80	MicroRNA Metabolism and Dysregulation in Amyotrophic Lateral Sclerosis. Molecular Neurobiology, 2018, 55, 2617-2630.	1.9	51
81	Subclinical Leberâ€™s hereditary optic neuropathy with pediatric acute spinal cord onset: more than meets the eye. BMC Neurology, 2018, 18, 220.	0.8	3
82	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.	1.1	6
83	Mitochondrial dysfunction in fibroblasts of Multiple System Atrophy. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 3588-3597.	1.8	32
84	Mitochondrial Dysregulation and Impaired Autophagy in iPSC-Derived Dopaminergic Neurons of Multiple System Atrophy. Stem Cell Reports, 2018, 11, 1185-1198.	2.3	46
85	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.	1.1	20
86	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.	1.1	6
87	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.	1.8	4
88	MicroRNA expression analysis identifies a subset of downregulated miRNAs in ALS motor neuron progenitors. Scientific Reports, 2018, 8, 10105.	1.6	53
89	Investigation of New Morpholino Oligomers to Increase Survival Motor Neuron Protein Levels in Spinal Muscular Atrophy. International Journal of Molecular Sciences, 2018, 19, 167.	1.8	8
90	Purkinje cell COX deficiency and mtDNA depletion in an animal model of spinocerebellar ataxia type 1. Journal of Neuroscience Research, 2018, 96, 1576-1585.	1.3	12

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91	The italian limb girdle muscular dystrophy registry: Relative frequency, clinical features, and differential diagnosis. Muscle and Nerve, 2017, 55, 55-68.	1.0	86
92	Cellular Therapy for Spinal Muscular Atrophy. , 2017, , 251-275.		0
93	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.	1.6	22
94	Mutations in TMEM230 are rare in autosomal dominant Parkinson's disease. Parkinsonism and Related Disorders, 2017, 39, 87-88.	1.1	11
95	MicroRNA-Directed Neuronal Reprogramming as a Therapeutic Strategy for Neurological Diseases. Molecular Neurobiology, 2017, 55, 4428-4436.	1.9	10
96	Development of Therapeutics for C9ORF72 ALS/FTD-Related Disorders. Molecular Neurobiology, 2017, 54, 4466-4476.	1.9	30
97	Morpholino-mediated SOD1 reduction ameliorates an amyotrophic lateral sclerosis disease phenotype. Scientific Reports, 2016, 6, 21301.	1.6	26
98	Differential neuronal vulnerability identifies IGF-2 as a protective factor in ALS. Scientific Reports, 2016, 6, 25960.	1.6	80
99	New Mutations in NEB Gene Discovered by Targeted Next-Generation Sequencing in Nemaline Myopathy Italian Patients. Journal of Molecular Neuroscience, 2016, 59, 351-359.	1.1	17
100	Mutational analysis of COQ2 in patients with MSA in Italy. Neurobiology of Aging, 2016, 45, 213.e1-213.e2.	1.5	25
101	Selective mitochondrial depletion, apoptosis resistance, and increased mitophagy in human Charcot-Marie-Tooth 2A motor neurons. Human Molecular Genetics, 2016, 25, 4266-4281.	1.4	41
102	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.	1.4	27
103	Clinical Pregenetic Screening for Stroke Monogenic Diseases. Stroke, 2016, 47, 1702-1709.	1.0	34
104	Autophagy in motor neuron disease: Key pathogenetic mechanisms and therapeutic targets. Molecular and Cellular Neurosciences, 2016, 72, 84-90.	1.0	43
105	Is spinal muscular atrophy a disease of the motor neurons only: pathogenesis and therapeutic implications?. Cellular and Molecular Life Sciences, 2016, 73, 1003-1020.	2.4	49
106	Experimental Advances Towards Neural Regeneration from Induced Stem Cells to Direct In Vivo Reprogramming. Molecular Neurobiology, 2016, 53, 2124-2131.	1.9	11
107	Clinical and molecular features and therapeutic perspectives of spinal muscular atrophy with respiratory distress type 1. Journal of Cellular and Molecular Medicine, 2015, 19, 2058-2066.	1.6	14
108	Spinal muscular atrophy phenotype is ameliorated in human motor neurons by SMN increase via different novel RNA therapeutic approaches. Scientific Reports, 2015, 5, 11746.	1.6	37

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109	ISPD mutations account for a small proportion of Italian Limb Girdle Muscular Dystrophy cases. BMC Neurology, 2015, 15, 172.	0.8	10
110	SOD1 misplacing and mitochondrial dysfunction in amyotrophic lateral sclerosis pathogenesis. Frontiers in Cellular Neuroscience, 2015, 9, 336.	1.8	111
111	Spinal muscular atrophyâ€"recent therapeutic advances for an old challenge. Nature Reviews Neurology, 2015, 11, 351-359.	4.9	119
112	Motor neurons with differential vulnerability to degeneration show distinct protein signatures in health and ALS. Neuroscience, 2015, 291, 216-229.	1.1	62
113	Gene therapy rescues disease phenotype in a spinal muscular atrophy with respiratory distress type 1 (SMARD1) mouse model. Science Advances, 2015, 1, e1500078.	4.7	33
114	Therapeutic Development in Amyotrophic Lateral Sclerosis. Clinical Therapeutics, 2015, 37, 668-680.	1.1	71
115	Novel Splice-Site Mutation in SMN1 Associated with a very Severe SMA-I Phenotype. Journal of Molecular Neuroscience, 2015, 56, 212-215.	1.1	11
116	Impaired Muscle Mitochondrial Biogenesis and Myogenesis in Spinal Muscular Atrophy. JAMA Neurology, 2015, 72, 666.	4.5	106
117	MFN2-related neuropathies: Clinical features, molecular pathogenesis and therapeutic perspectives. Journal of the Neurological Sciences, 2015, 356, 7-18.	0.3	112
118	A novel homozygous PLA2G6 mutation causes dystonia-parkinsonism. Parkinsonism and Related Disorders, 2015, 21, 337-339.	1.1	22
119	TUBA4A gene analysis in sporadic amyotrophic lateral sclerosis: identification of novel mutations. Journal of Neurology, 2015, 262, 1376-1378.	1.8	44
120	Human pluripotent stem cells as tools for neurodegenerative and neurodevelopmental disease modeling and drug discovery. Expert Opinion on Drug Discovery, 2015, 10, 615-629.	2.5	49
121	CHCHD10mutations in Italian patients with sporadic amyotrophic lateral sclerosis: Figure 1. Brain, 2015, 138, e372-e372.	3.7	59
122	Pluripotent stem cell-based models of spinal muscular atrophy. Molecular and Cellular Neurosciences, 2015, 64, 44-50.	1.0	28
123	Therapeutic applications of the cell-penetrating HIV-1 Tat peptide. Drug Discovery Today, 2015, 20, 76-85.	3.2	173
124	iPSC-Based Models to Unravel Key Pathogenetic Processes Underlying Motor Neuron Disease Development. Journal of Clinical Medicine, 2014, 3, 1124-1145.	1.0	6
125	Lower motor neuron disease with respiratory failure caused by a novel MAPT mutation. Neurology, 2014, 82, 1990-1998.	1.5	21
126	Minimally invasive transplantation of iPSC-derived ALDHhiSSCloVLA4+ neural stem cells effectively improves the phenotype of an amyotrophic lateral sclerosis model. Human Molecular Genetics, 2014, 23, 342-354.	1.4	97

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127	Analysis of the KIFAP3 gene in amyotrophic lateral sclerosis: a multicenter survival study. Neurobiology of Aging, 2014, 35, 2420.e13-2420.e14.	1.5	16
128	In vitro neurogenesis: development and functional implications of iPSC technology. Cellular and Molecular Life Sciences, 2014, 71, 1623-1639.	2.4	39
129	Stem cell transplantation for amyotrophic lateral sclerosis: therapeutic potential and perspectives on clinical translation. Cellular and Molecular Life Sciences, 2014, 71, 3257-3268.	2.4	32
130	Cellular therapy to target neuroinflammation in amyotrophic lateral sclerosis. Cellular and Molecular Life Sciences, 2014, 71, 999-1015.	2.4	89
131	Effect of Combined Systemic and Local Morpholino Treatment on the Spinal Muscular Atrophy Î”7 Mouse Model Phenotype. Clinical Therapeutics, 2014, 36, 340-356.e5.	1.1	44
132	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.	1.6	20
133	Antisense Oligonucleotide Therapy for the Treatment of C9ORF72 ALS/FTD Diseases. Molecular Neurobiology, 2014, 50, 721-732.	1.9	48
134	Molecular Therapeutic Strategies for Spinal Muscular Atrophies: Current and Future Clinical Trials. Clinical Therapeutics, 2014, 36, 128-140.	1.1	74
135	A genome-wide association meta-analysis identifies a novel locus at 17q11.2 associated with sporadic amyotrophic lateral sclerosis. Human Molecular Genetics, 2014, 23, 2220-2231.	1.4	123
136	Induced neural stem cells: Methods of reprogramming and potential therapeutic applications. Progress in Neurobiology, 2014, 114, 15-24.	2.8	39
137	The wide spectrum of clinical phenotypes of spinal muscular atrophy with respiratory distress type 1: A systematic review. Journal of the Neurological Sciences, 2014, 346, 35-42.	0.3	30
138	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. Neuron, 2014, 84, 324-331.	3.8	308
139	Motor neuron derivation from human embryonic and induced pluripotent stem cells: experimental approaches and clinical perspectives. Stem Cell Research and Therapy, 2014, 5, 87.	2.4	52
140	Glycogen storage disease type III: A novel Agl knockout mouse model. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2014, 1842, 2318-2328.	1.8	28
141	iPSC-Derived Neural Stem Cells Act via Kinase Inhibition to Exert Neuroprotective Effects in Spinal Muscular Atrophy with Respiratory DistressÂType 1. Stem Cell Reports, 2014, 3, 297-311.	2.3	34
142	A novel CCM1mutation associated with multiple cerebral and vertebral cavernous malformations. BMC Neurology, 2014, 14, 158.	0.8	12
143	In vitro analysis of splice site mutations in the CLCN1 gene using the minigene assay. Molecular Biology Reports, 2014, 41, 2865-2874.	1.0	8
144	Ongoing therapeutic trials and outcome measures for Duchenne muscular dystrophy. Cellular and Molecular Life Sciences, 2013, 70, 4585-4602.	2.4	53

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145	POLG1 mutations and stroke like episodes: a distinct clinical entity rather than an atypical MELAS syndrome. BMC Neurology, 2013, 13, 8.	0.8	26
146	Direct Reprogramming of Adult Somatic Cells into other Lineages: Past Evidence and Future Perspectives. Cell Transplantation, 2013, 22, 921-944.	1.2	20
147	Mutations in DNA2 Link Progressive Myopathy to Mitochondrial DNA Instability. American Journal of Human Genetics, 2013, 92, 293-300.	2.6	115
148	Screening of the PFN1 gene in sporadic amyotrophic lateral sclerosis and in frontotemporal dementia. Neurobiology of Aging, 2013, 34, 1517.e9-1517.e10.	1.5	35
149	Analysis of hnRNPA1, A2/B1, and A3 genes in patients with amyotrophic lateral sclerosis. Neurobiology of Aging, 2013, 34, 2695.e11-2695.e12.	1.5	30
150	Mitochondrial Fusion Proteins and Human Diseases. Neurology Research International, 2013, 2013, 1-11.	0.5	85
151	Ubiquilin 2mutations in Italian patients with amyotrophic lateral sclerosis and frontotemporal dementia. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 183-187.	0.9	74
152	Riboflavin transporter 3 involvement in infantile Brown-Vialetto-Van Laere disease: two novel mutations. Journal of Medical Genetics, 2013, 50, 104-107.	1.5	31
153	Growing Evidence about the Relationship between Vessel Dissection and Scuba Diving. Case Reports in Neurology, 2013, 5, 155-161.	0.3	7
154	The novel mitochondrial tRNAAsn gene mutation m.5709T>C produces ophthalmoparesis and respiratory impairment. European Journal of Human Genetics, 2012, 20, 357-360.	1.4	4
155	Next-generation sequencing reveals DGUOK mutations in adult patients with mitochondrial DNA multiple deletions. Brain, 2012, 135, 3404-3415.	3.7	81
156	Genetic Correction of Human Induced Pluripotent Stem Cells from Patients with Spinal Muscular Atrophy. Science Translational Medicine, 2012, 4, 165ra162.	5.8	180
157	C9ORF72 repeat expansion in a large Italian ALS cohort: evidence of a founder effect. Neurobiology of Aging, 2012, 33, 2528.e7-2528.e14.	1.5	74
158	Generation of skeletal muscle cells from embryonic and induced pluripotent stem cells as an in vitro model and for therapy of muscular dystrophies. Journal of Cellular and Molecular Medicine, 2012, 16, 1353-1364.	1.6	61
159	Frequency and characterisation of anoctamin 5 mutations in a cohort of Italian limb-girdle muscular dystrophy patients. Neuromuscular Disorders, 2012, 22, 934-943.	0.3	53
160	Optic atrophy plus phenotype due to mutations in the OPA1 gene: Two more Italian families. Journal of the Neurological Sciences, 2012, 315, 146-149.	0.3	21
161	Myotonia congenita: Novel mutations in CLCN1 gene and functional characterizations in Italian patients. Journal of the Neurological Sciences, 2012, 318, 65-71.	0.3	22
162	Research advances in gene therapy approaches for the treatment of amyotrophic lateral sclerosis. Cellular and Molecular Life Sciences, 2012, 69, 1641-1650.	2.4	19

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163	Direct reprogramming of human astrocytes into neural stem cells and neurons. Experimental Cell Research, 2012, 318, 1528-1541.	1.2	143
164	New molecular findings in congenital myopathies due to selenoprotein N gene mutations. Journal of the Neurological Sciences, 2011, 300, 107-113.	0.3	23
165	Intracerebral haemorrhage, a possible presentation in Churg-Strauss syndrome: Case report and review of the literature. Journal of the Neurological Sciences, 2011, 301, 107-111.	0.3	19
166	Two novel mutations in PEO1 (Twinkle) gene associated with chronic external ophthalmoplegia. Journal of the Neurological Sciences, 2011, 308, 173-176.	0.3	7
167	Unusual adult-onset Leigh syndrome presentation due to the mitochondrial m.9176T>C mutation. Biochemical and Biophysical Research Communications, 2011, 412, 245-248.	1.0	19
168	ALS genetic modifiers that increase survival of SOD1 mice and are suitable for therapeutic development. Progress in Neurobiology, 2011, 95, 133-148.	2.8	26
169	No major progranulin genetic variability contribution to disease etiopathogenesis in an ALS Italian cohort. Neurobiology of Aging, 2011, 32, 1157-1158.	1.5	18
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