Giacomo P Comi

List of Publications by Year
in descending order

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		9756	20307
459	20,885	73	116
papers	citations	h-index	g-index

468	468	468	23342
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Spinal muscular atrophy: state of the art and new therapeutic strategies. Neurological Sciences, 2022, 43, 615-624.	0.9	13
2	Cell-penetrating peptide-conjugated Morpholino rescues SMA in a symptomatic preclinical model. Molecular Therapy, 2022, 30, 1288-1299.	3.7	12
3	TM6SF2/PNPLA3/MBOAT7 Loss-of-Function Genetic Variants Impact on NAFLD Development and Progression Both in Patients and in InÂVitro Models. Cellular and Molecular Gastroenterology and Hepatology, 2022, 13, 759-788.	2.3	44
4	VPS13C-associated Parkinson's disease: Two novel cases and review of the literature. Parkinsonism and Related Disorders, 2022, 94, 37-39.	1.1	8
5	Mitochondrial DNA homeostasis impairment and dopaminergic dysfunction: A trembling balance. Ageing Research Reviews, 2022, 76, 101578.	5.0	15
6	Safety and efficacy of once-daily risdiplam in type 2 and non-ambulant type 3 spinal muscular atrophy (SUNFISH part 2): a phase 3, double-blind, randomised, placebo-controlled trial. Lancet Neurology, The, 2022, 21, 42-52.	4.9	89
7	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
8	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
9	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
10	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
11	Stathmins and Motor Neuron Diseases: Pathophysiology and Therapeutic Targets. Biomedicines, 2022, 10, 711.	1.4	9
12	Biallelic Variants in ENDOG Associated with Mitochondrial Myopathy and Multiple mtDNA Deletions. Cells, 2022, 11, 974.	1.8	4
13	Muscle histological changes in a large cohort of patients affected with Becker muscular dystrophy. Acta Neuropathologica Communications, 2022, 10, 48.	2.4	11
14	Characterization of patients with Becker muscular dystrophy by histology, magnetic resonance imaging, function, and strength assessments. Muscle and Nerve, 2022, 65, 326-333.	1.0	8
15	Homozygous SOD1 Variation L144S Produces a Severe Form of Amyotrophic Lateral Sclerosis in an Iranian Family. Neurology: Genetics, 2022, 8, e645.	0.9	6
16	Clinical and genetic features of a cohort of patients with MFN2-related neuropathy. Scientific Reports, 2022, 12, 6181.	1.6	10
17	Genetic modifiers of upper limb function in Duchenne muscular dystrophy. Journal of Neurology, 2022, 269, 4884-4894.	1.8	2
18	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

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19	A novel RRM2B mutation associated with mitochondrial DNA depletion syndrome. Molecular Genetics and Metabolism Reports, 2022, 32, 100887.	0.4	2
20	New Insights into Cerebral Vessel Disease Landscapes at Single-Cell Resolution: Pathogenetic and Therapeutic Perspectives. Biomedicines, 2022, 10, 1693.	1.4	1
21	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
22	Expanding the genotypic and phenotypic spectrum of Betaâ€propeller poteinâ€associated neurodegeneration. European Journal of Neurology, 2021, 28, e25-e27.	1.7	0
23	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
24	A Novel Homozygous <scp>VPS11</scp> Variant May Cause Generalized Dystonia. Annals of Neurology, 2021, 89, 834-839.	2.8	13
25	Clinical features and disease course of patients with acute ischaemic stroke just before the Italian index case: Was COVID-19 already there?. Internal and Emergency Medicine, 2021, 16, 1247-1252.	1.0	0
26	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
27	Advancing Drug Discovery for Neurological Disorders Using iPSC-Derived Neural Organoids. International Journal of Molecular Sciences, 2021, 22, 2659.	1.8	33
28	Missense mutations in small muscle protein X-linked (SMPX) cause distal myopathy with protein inclusions. Acta Neuropathologica, 2021, 142, 375-393.	3.9	6
29	Charcotâ€“Marieâ€“Tooth disease type 2F associated with biallelic HSPB1 mutations. Annals of Clinical and Translational Neurology, 2021, 8, 1158-1164.	1.7	4
30	Early Findings in Neonatal Cases of RYR1â€“Related Congenital Myopathies. Frontiers in Neurology, 2021, 12, 664618.	1.1	3
31	North Star Ambulatory Assessment changes in ambulant Duchenne boys amenable to skip exons 44, 45, 51, and 53: A 3 year follow up. PLoS ONE, 2021, 16, e0253882.	1.1	6
32	The nonsense mutation stop+4 model correlates with motor changes in Duchenne muscular dystrophy. Neuromuscular Disorders, 2021, 31, 479-488.	0.3	0
33	Massive cerebral venous thrombosis due to vaccine-induced immune thrombotic thrombocytopenia. Haematologica, 2021, 106, 3021-3024.	1.7	8
34	Co-occurrence of DMPK expansion and CLCN1 mutation in a patient with myotonia. Neurological Sciences, 2021, 42, 5365-5368.	0.9	2
35	p.Asn1180Ile mutation of SCN4A gene in an Italian family with myopathy and myotonic syndrome. Neurological Sciences, 2021, 42, 5359-5363.	0.9	0
36	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

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37	Clinical, neuroradiological and genetic findings in a cohort of patients with multiple Cerebral Cavernous Malformations. Metabolic Brain Disease, 2021, 36, 1871-1878.	1.4	5
38	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. JAMA Neurology, 2021, 78, 1236.	4.5	46
39	Screening of LRP10 mutations in Parkinson's disease patients from Italy. Parkinsonism and Related Disorders, 2021, 89, 17-21.	1.1	5
40	Perspectives on hiPSC-Derived Muscle Cells as Drug Discovery Models for Muscular Dystrophies. International Journal of Molecular Sciences, 2021, 22, 9630.	1.8	3
41	Sodium Levels Predict Disability at Discharge in Guillain-BarrÃ© Syndrome: A Retrospective Cohort Study. Frontiers in Neurology, 2021, 12, 729252.	1.1	2
42	Starting HIV therapy in patients with mitochondrial disease. Aids, 2021, 35, 2063-2065.	1.0	0
43	Consumption of complement in a 26-year-old woman with severe thrombotic thrombocytopenia after ChAdOx1 nCov-19 vaccination. Journal of Autoimmunity, 2021, 124, 102728.	3.0	5
44	Fluency type index: A neuropsychological marker to predict amnestic mild cognitive impairment progression to Alzheimer's disease. Journal of the Neurological Sciences, 2021, 429, 119005.	0.3	0
45	Human spinal cord-like organoids to model C9ORF72 ALS and test new therapies in vitro. Journal of the Neurological Sciences, 2021, 429, 117761.	0.3	0
46	A young male with walking difficulties and subacute brainstem dysfunction: Adult-onset Leigh syndrome. Journal of the Neurological Sciences, 2021, 429, 119363.	0.3	0
47	CACNA1S mutation associated with a case of juvenile-onset congenital myopathy. Journal of the Neurological Sciences, 2021, 431, 120047.	0.3	2
48	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
49	Molecular Approaches for the Treatment of Pompe Disease. Molecular Neurobiology, 2020, 57, 1259-1280.	1.9	17
50	Anti-MAG IgM: differences in antibody tests and correlation with clinical findings. Neurological Sciences, 2020, 41, 365-372.	0.9	9
51	MRI patterns of muscle involvement in type 2 and 3 spinal muscular atrophy patients. Journal of Neurology, 2020, 267, 898-912.	1.8	32
52	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
53	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
54	Expanding the clinical spectrum of the mitochondrial mutation A13084T in the ND5 gene. Neurology: Genetics, 2020, 6, e511.	0.9	1

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55	Dystoniaâ€ataxia syndrome with permanent torsional nystagmus caused by ECHS1 deficiency. Annals of Clinical and Translational Neurology, 2020, 7, 839-845.	1.7	10
56	Muscle MRI in two SMA patients on nusinersen treatment: A two years follow-up. Journal of the Neurological Sciences, 2020, 417, 117067.	0.3	13
57	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
58	New genotype-phenotype correlations in a large European cohort of patients with sarcoglycanopathy. Brain, 2020, 143, 2696-2708.	3.7	45
59	Insights into disease mechanisms and potential therapeutics for C9orf72-related amyotrophic lateral sclerosis/frontotemporal dementia. Ageing Research Reviews, 2020, 64, 101172.	5.0	5
60	Animal Models of CMT2A: State-of-art and Therapeutic Implications. Molecular Neurobiology, 2020, 57, 5121-5129.	1.9	6
61	Meta-analyses of ataluren randomized controlled trials in nonsense mutation Duchenne muscular dystrophy. Journal of Comparative Effectiveness Research, 2020, 9, 973-984.	0.6	41
62	TYMP Variants Result in Late-Onset Mitochondrial Myopathy With Altered Muscle Mitochondrial DNA Homeostasis. Frontiers in Genetics, 2020, 11, 860.	1.1	6
63	Hereditary hemorrhagic telangiectasia associated with cortical development malformation due to a start loss mutation in ENG. BMC Neurology, 2020, 20, 316.	0.8	8
64	Late-onset leukoencephalopathy in a patient with recessive EARS2 mutations. Neurology: Genetics, 2020, 6, e488.	0.9	0
65	Primary mitochondrial myopathy. Neurology: Genetics, 2020, 6, e519.	0.9	10
66	Long-term follow-up of patients with type 2 and non-ambulant type 3 spinal muscular atrophy (SMA) treated with olesoxime in the OLEOS trial. Neuromuscular Disorders, 2020, 30, 959-969.	0.3	15
67	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
68	Pediatric anti-HMGCR necrotizing myopathy: diagnostic challenges and literature review. Neurological Sciences, 2020, 41, 3009-3013.	0.9	6
69	The Genetic Landscape of Dystrophin Mutations in Italy: A Nationwide Study. Frontiers in Genetics, 2020, 11, 131.	1.1	49
70	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
71	MYH2 myopathy, a new case expands the clinical and pathological spectrum of the recessive form. Molecular Genetics & Genomic Medicine, 2020, 8, e1320.	0.6	10
72	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

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73	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
74	Genetic modifiers of respiratory function in Duchenne muscular dystrophy. Annals of Clinical and Translational Neurology, 2020, 7, 786-798.	1.7	36
75	Sodium Channel Myotonia Due to Novel Mutations in Domain I of Nav1.4. Frontiers in Neurology, 2020, 11, 255.	1.1	5
76	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
77	Neural Stem Cell Transplantation for Neurodegenerative Diseases. International Journal of Molecular Sciences, 2020, 21, 3103.	1.8	105
78	Glial cells involvement in spinal muscular atrophy: Could SMA be a neuroinflammatory disease?. Neurobiology of Disease, 2020, 140, 104870.	2.1	35
79	SLC25A46 mutations in patients with Parkinson's Disease and optic atrophy. Parkinsonism and Related Disorders, 2020, 74, 1-5.	1.1	16
80	The Role of Mitochondria in Neurodegenerative Diseases: the Lesson from Alzheimerâ€™s Disease and Parkinsonâ€™s Disease. Molecular Neurobiology, 2020, 57, 2959-2980.	1.9	180
81	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
82	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
83	R-Loops in Motor Neuron Diseases. Molecular Neurobiology, 2019, 56, 2579-2589.	1.9	39
84	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
85	Novel mutations in DNA2 associated with myopathy and mtDNA instability. Annals of Clinical and Translational Neurology, 2019, 6, 1893-1899.	1.7	13
86	Loss of the nucleoporin Aladin in central nervous system and fibroblasts of Allgrove Syndrome. Human Molecular Genetics, 2019, 28, 3921-3927.	1.4	9
87	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
88	Ophthalmoplegia Due to Miller Fisher Syndrome in a Patient With Myasthenia Gravis. Frontiers in Neurology, 2019, 10, 823.	1.1	4
89	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
90	Muscle pain in mitochondrial diseases: a picture from the Italian network. Journal of Neurology, 2019, 266, 953-959.	1.8	9

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91	Long-term natural history data in Duchenne muscular dystrophy ambulant patients with mutations amenable to skip exons 44, 45, 51 and 53. PLoS ONE, 2019, 14, e0218683.	1.1	47
92	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
93	Disease Modeling and Therapeutic Strategies in CMT2A: State of the Art. Molecular Neurobiology, 2019, 56, 6460-6471.	1.9	20
94	Neurofascin (NFASC) gene mutation causes autosomal recessive ataxia with demyelinating neuropathy. Parkinsonism and Related Disorders, 2019, 63, 66-72.	1.1	25
95	Advances, Challenges, and Perspectives in Translational Stem Cell Therapy for Amyotrophic Lateral Sclerosis. Molecular Neurobiology, 2019, 56, 6703-6715.	1.9	24
96	Lipomatosis Incidence and Characteristics in an Italian Cohort of Mitochondrial Patients. Frontiers in Neurology, 2019, 10, 160.	1.1	19
97	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
98	Cardiac and Neuromuscular Features of Patients WithLMNA-Related Cardiomyopathy. Annals of Internal Medicine, 2019, 171, 458.	2.0	33
99	Elucidating the role of Agl in bladder carcinogenesis by generation and characterization of genetically engineered mice. Carcinogenesis, 2019, 40, 194-201.	1.3	2
100	Fibrosis Rescue Improves Cardiac Function in Dystrophin-Deficient Mice and Duchenne Patientâ€“Specific Cardiomyocytes by Immunoproteasome Modulation. American Journal of Pathology, 2019, 189, 339-353.	1.9	27
101	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
102	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
103	Targeted gene panel screening is an effective tool to identify undiagnosed late onset Pompe disease. Neuromuscular Disorders, 2018, 28, 586-591.	0.3	24
104	Interpreting Genetic Variants in Titin in Patients With Muscle Disorders. JAMA Neurology, 2018, 75, 557.	4.5	69
105	Reply: DGUOK recessive mutations in patients with CPEO, mitochondrial myopathy, parkinsonism and mtDNA deletions. Brain, 2018, 141, e4-e4.	3.7	3
106	Rescue of GSDIII Phenotype with Gene Transfer Requires Liver- and Muscle-Targeted GDE Expression. Molecular Therapy, 2018, 26, 890-901.	3.7	24
107	A de novo C19orf12 heterozygous mutation in a patient with MPAN. Parkinsonism and Related Disorders, 2018, 48, 109-111.	1.1	15
108	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

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109	Novel Lys215Asn mutation in an Italian family with Thomsen myotonia. Neurological Sciences, 2018, 39, 1491-1492.	0.9	1
110	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	3.8	517
111	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
112	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
113	Mitochondrial dysfunction in fibroblasts of Multiple System Atrophy. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 3588-3597.	1.8	32
114	Mitochondrial Dysregulation and Impaired Autophagy in iPSC-Derived Dopaminergic Neurons of Multiple System Atrophy. Stem Cell Reports, 2018, 11, 1185-1198.	2.3	46
115	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
116	Copy Number Variants Account for a Tiny Fraction of Undiagnosed Myopathic Patients. Genes, 2018, 9, 524.	1.0	7
117	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
118	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.	0.3	11
119	Rhabdomyolysis-Associated Acute Kidney Injury. American Journal of Kidney Diseases, 2018, 71, A12-A14.	2.1	26
120	The analysis of myotonia congenita mutations discloses functional clusters of amino acids within the CBS2 domain and the C-terminal peptide of the ClC-1 channel. Human Mutation, 2018, 39, 1273-1283.	1.1	15
121	The MELAS mutation m.3243A>G promotes reactivation of fetal cardiac genes and an epithelial-mesenchymal transition-like program via dysregulation of miRNAs. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 3022-3037.	1.8	12
122	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
123	Secondary prevention of cryptogenic stroke in patients with patent foramen ovale: a systematic review and meta-analysis. Internal and Emergency Medicine, 2018, 13, 1287-1303.	1.0	3
124	The Length of SNCA Rep1 Microsatellite May Influence Cognitive Evolution in Parkinsonâ€™s Disease. Frontiers in Neurology, 2018, 9, 213.	1.1	21
125	MicroRNA expression analysis identifies a subset of downregulated miRNAs in ALS motor neuron progenitors. Scientific Reports, 2018, 8, 10105.	1.6	53
126	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

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127	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
128	The italian limb girdle muscular dystrophy registry: Relative frequency, clinical features, and differential diagnosis. Muscle and Nerve, 2017, 55, 55-68.	1.0	86
129	Diagnosis of Duchenne Muscular Dystrophy in Italy in the last decade: Critical issues and areas for improvements. Neuromuscular Disorders, 2017, 27, 447-451.	0.3	42
130	Neuromuscular excitability changes produced by sustained voluntary contraction and response to mexiletine in myotonia congenita. Neurophysiologie Clinique, 2017, 47, 247-252.	1.0	11
131	Safety and efficacy of olesoxime in patients with type 2 or non-ambulatory type 3 spinal muscular atrophy: a randomised, double-blind, placebo-controlled phase 2 trial. Lancet Neurology, The, 2017, 16, 513-522.	4.9	95
132	Anti-sulfatide reactivity in patients with celiac disease. Scandinavian Journal of Gastroenterology, 2017, 52, 409-413.	0.6	2
133	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
134	Mutations in TMEM230 are rare in autosomal dominant Parkinson's disease. Parkinsonism and Related Disorders, 2017, 39, 87-88.	1.1	11
135	Biallelic C1QBP Mutations Cause Severe Neonatal-, Childhood-, or Later-Onset Cardiomyopathy Associated with Combined Respiratory-Chain Deficiencies. American Journal of Human Genetics, 2017, 101, 525-538.	2.6	58
136	Development of Therapeutics for C9ORF72 ALS/FTD-Related Disorders. Molecular Neurobiology, 2017, 54, 4466-4476.	1.9	30
137	LOPED study: looking for an early diagnosis in a late-onset Pompe disease high-risk population. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, jnnp-2014-310164.	0.9	50
138	Timed Rise from Floor as a Predictor of Disease Progression in Duchenne Muscular Dystrophy: An Observational Study. PLoS ONE, 2016, 11, e0151445.	1.1	32
139	Morpholino-mediated SOD1 reduction ameliorates an amyotrophic lateral sclerosis disease phenotype. Scientific Reports, 2016, 6, 21301.	1.6	26
140	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
141	Registries versus tertiary care centers: How do we measure standards of care in Duchenne muscular dystrophy?. Neuromuscular Disorders, 2016, 26, 261-263.	0.3	3
142	Histological effects of givinostat in boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 2016, 26, 643-649.	0.3	144
143	Mutational analysis of COQ2 in patients with MSA in Italy. Neurobiology of Aging, 2016, 45, 213.e1-213.e2.	1.5	25
144	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

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145	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1043-1048.	9.4	494
146	Asymptomatic Pompe disease: Can muscle magnetic resonance imaging facilitate diagnosis?. Muscle and Nerve, 2016, 53, 326-327.	1.0	7
147	The genetic basis of undiagnosed muscular dystrophies and myopathies. Neurology, 2016, 87, 71-76.	1.5	92
148	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
149	Clinical Pregenetic Screening for Stroke Monogenic Diseases. Stroke, 2016, 47, 1702-1709.	1.0	34
150	Association of a Locus in theCAMTA1Gene With Survival in Patients With Sporadic Amyotrophic Lateral Sclerosis. JAMA Neurology, 2016, 73, 812.	4.5	57
151	Response to: Mitochondrial neuropathy affects peripheral and cranial nerves and is primary or secondary or both. Neuromuscular Disorders, 2016, 26, 549.	0.3	0
152	â€œMitochondrial neuropathiesâ€: A survey from the large cohort of the Italian Network. Neuromuscular Disorders, 2016, 26, 272-276.	0.3	37
153	Loss of Glycogen Debranching Enzyme AGL Drives Bladder Tumor Growth via Induction of Hyaluronic Acid Synthesis. Clinical Cancer Research, 2016, 22, 1274-1283.	3.2	25
154	Longitudinal follow-up and muscle MRI pattern of two siblings with polyglucosan body myopathy due to glycogenin-1 mutation. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 797-800.	0.9	17
155	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
156	ISPD mutations account for a small proportion of Italian Limb Girdle Muscular Dystrophy cases. BMC Neurology, 2015, 15, 172.	0.8	10
157	Genes and Pathways Involved in Adult Onset Disorders Featuring Muscle Mitochondrial DNA Instability. International Journal of Molecular Sciences, 2015, 16, 18054-18076.	1.8	25
158	SOD1 misplacing and mitochondrial dysfunction in amyotrophic lateral sclerosis pathogenesis. Frontiers in Cellular Neuroscience, 2015, 9, 336.	1.8	111
159	Improvement of Endurance of DMD Animal Model Using Natural Polyphenols. BioMed Research International, 2015, 2015, 1-17.	0.9	11
160	Spinal muscular atrophyâ€"recent therapeutic advances for an old challenge. Nature Reviews Neurology, 2015, 11, 351-359.	4.9	119
161	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
162	Prevalence of congenital muscular dystrophy in Italy. Neurology, 2015, 84, 904-911.	1.5	75

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163	Platelet mitochondrial dysfunction in critically ill patients: comparison between sepsis and cardiogenic shock. Critical Care, 2015, 19, 39.	2.5	41
164	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
165	Impaired Muscle Mitochondrial Biogenesis and Myogenesis in Spinal Muscular Atrophy. JAMA Neurology, 2015, 72, 666.	4.5	106
166	MFN2-related neuropathies: Clinical features, molecular pathogenesis and therapeutic perspectives. Journal of the Neurological Sciences, 2015, 356, 7-18.	0.3	112
167	A novel homozygous PLA2G6 mutation causes dystonia-parkinsonism. Parkinsonism and Related Disorders, 2015, 21, 337-339.	1.1	22
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