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	Human mitochondrial DNA deletions associated with mutations in the gene encoding Twinkle, a phage T7 gene 4-like protein localized in mitochondria. Nature Genetics, 2001, 28, 223-231.	9.4	803
2	Role of Adenine Nucleotide Translocator 1 in mtDNA Maintenance. Science, 2000, 289, 782-785.	6.0	591
3	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	3.8	517
4	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
5	Ataluren treatment of patients with nonsense mutation dystrophinopathy. Muscle and Nerve, 2014, 50, 477-487.	1.0	357
6	Mutations of mitochondrial DNA polymerase ?A are a frequent cause of autosomal dominant or recessive progressive external ophthalmoplegia. Annals of Neurology, 2002, 52, 211-219.	2.8	257
7	Cytochromec Oxidase subunit I microdeletion in a patient with motor neuron disease. Annals of Neurology, 1998, 43, 110-116.	2.8	251
8	Identification of a Primitive Brain-Derived Neural Stem Cell Population Based on Aldehyde Dehydrogenase Activity. Stem Cells, 2006, 24, 975-985.	1.4	240
9	Parasites represent a major selective force for interleukin genes and shape the genetic predisposition to autoimmune conditions. Journal of Experimental Medicine, 2009, 206, 1395-1408.	4.2	230
10	Polyneuropathy in POEMS syndrome: role of angiogenic factors in the pathogenesis. Brain, 2005, 128, 1911-1920.	3.7	216
11	Autologous Transplantation of Muscle-Derived CD133+ Stem Cells in Duchenne Muscle Patients. Cell Transplantation, 2007, 16, 563-577.	1.2	214
12	Loss-of-function mutations in MGME1 impair mtDNA replication and cause multisystemic mitochondrial disease. Nature Genetics, 2013, 45, 214-219.	9.4	198
13	Wild-type bone marrow cells ameliorate the phenotype of SOD1-G93A ALS mice and contribute to CNS, heart and skeletal muscle tissues. Brain, 2004, 127, 2518-2532.	3.7	187
14	Genetic Correction of Human Induced Pluripotent Stem Cells from Patients with Spinal Muscular Atrophy. Science Translational Medicine, 2012, 4, 165ra162.	5.8	180
15	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
16	Infantile Encephalopathy and Defective Mitochondrial DNA Translation in Patients with Mutations of Mitochondrial Elongation Factors EFG1 and EFTu. American Journal of Human Genetics, 2007, 80, 44-58.	2.6	172
17	Reliability of the North Star Ambulatory Assessment in a multicentric setting. Neuromuscular Disorders, 2009, 19, 458-461.	0.3	171
18	Congenital muscular dystrophies with defective glycosylation of dystroglycan. Neurology, 2009, 72, 1802-1809.	1.5	166

#	Article	IF	Citations
19	Clinical, molecular, and protein correlations in a large sample of genetically diagnosed Italian limb girdle muscular dystrophy patients. Human Mutation, 2008, 29, 258-266.	1.1	162
20	Phenotypic heterogeneity of the 8344A>G mtDNA â€œMERRFâ€•mutation. Neurology, 2013, 80, 2049-2054.	1.5	157
21	Cognitive impairment in Duchenne muscular dystrophy. Neuromuscular Disorders, 1994, 4, 359-369.	0.3	152
22	Mutations of FUS gene in sporadic amyotrophic lateral sclerosis. Journal of Medical Genetics, 2010, 47, 190-194.	1.5	152
23	Widespread balancing selection and pathogen-driven selection at blood group antigen genes. Genome Research, 2009, 19, 199-212.	2.4	147
24	Histological effects of givinostat in boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 2016, 26, 643-649.	0.3	144
25	Direct reprogramming of human astrocytes into neural stem cells and neurons. Experimental Cell Research, 2012, 318, 1528-1541.	1.2	143
26	Identification of Novel Mutations in the Ryanodine-Receptor Gene (RYR1) in Malignant Hyperthermia: Genotype-Phenotype Correlation. American Journal of Human Genetics, 1998, 62, 599-609.	2.6	141
27	Genotype and phenotype characterization in a large dystrophinopathic cohort with extended follow-up. Journal of Neurology, 2011, 258, 1610-1623.	1.8	134
28	Neural stem cells LewisX + CXCR4 + modify disease progression in an amyotrophic lateral sclerosis model. Brain, 2007, 130, 1289-1305.	3.7	127
29	?-enolase deficiency, a new metabolic myopathy of distal glycolysis. Annals of Neurology, 2001, 50, 202-207.	2.8	125
30	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
31	Mitochondrial Respiratory Chain Dysfunction in Muscle From Patients With Amyotrophic Lateral Sclerosis. Archives of Neurology, 2010, 67, 849-54.	4.9	122
32	Loss-of-function mutations of SURF-1 are specifically associated with Leigh syndrome with cytochromec oxidase deficiency. Annals of Neurology, 1999, 46, 161-166.	2.8	121
33	The Mitochondrial Disulfide Relay System Protein GFER Is Mutated in Autosomal-Recessive Myopathy with Cataract and Combined Respiratory-Chain Deficiency. American Journal of Human Genetics, 2009, 84, 594-604.	2.6	121
34	Silencer elements as possible inhibitors of pseudoexon splicing. Nucleic Acids Research, 2004, 32, 1783-1791.	6.5	120
35	Neural stem cell transplantation can ameliorate the phenotype of a mouse model of spinal muscular atrophy. Journal of Clinical Investigation, 2008, 118, 3316-3330.	3.9	119
36	Stem cell therapy in stroke. Cellular and Molecular Life Sciences, 2009, 66, 757-772.	2.4	119

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37	The m.3243A>G mitochondrial DNA mutation and related phenotypes. A matter of gender?. Journal of Neurology, 2014, 261, 504-510.	1.8	119
38	Spinal muscular atrophyâ€"recent therapeutic advances for an old challenge. Nature Reviews Neurology, 2015, 11, 351-359.	4.9	119
39	Genotype to phenotype correlations in mitochondrial encephalomyopathies associated with the A3243G mutation of mitochondrial DNA. Journal of Neurology, 1995, 242, 304-312.	1.8	115
40	Vascular endothelial growth factor gene variability is associated with increased risk for AD. Annals of Neurology, 2005, 57, 373-380.	2.8	115
41	Mutations in DNA2 Link Progressive Myopathy to Mitochondrial DNA Instability. American Journal of Human Genetics, 2013, 92, 293-300.	2.6	115
42	MFN2-related neuropathies: Clinical features, molecular pathogenesis and therapeutic perspectives. Journal of the Neurological Sciences, 2015, 356, 7-18.	0.3	112
43	Mitochondrial defect and PGC-1Î± dysfunction in parkin-associated familial Parkinson's disease. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2011, 1812, 1041-1053.	1.8	111
44	SOD1 misplacing and mitochondrial dysfunction in amyotrophic lateral sclerosis pathogenesis. Frontiers in Cellular Neuroscience, 2015, 9, 336.	1.8	111
45	Impaired Muscle Mitochondrial Biogenesis and Myogenesis in Spinal Muscular Atrophy. JAMA Neurology, 2015, 72, 666.	4.5	106
46	Autosomal Dominant Frontotemporal Lobar Degeneration Due to the C9ORF72 Hexanucleotide Repeat Expansion: Late-Onset Psychotic Clinical Presentation. Biological Psychiatry, 2013, 74, 384-391.	0.7	105
47	Neural Stem Cell Transplantation for Neurodegenerative Diseases. International Journal of Molecular Sciences, 2020, 21, 3103.	1.8	105
48	Isolation and characterization of murine neural stem/progenitor cells based on Prominin-1 expression. Experimental Neurology, 2007, 205, 547-562.	2.0	104
49	Retrospective study of a large population of patients with asymptomatic or minimally symptomatic raised serum creatine kinase levels. Journal of Neurology, 2002, 249, 305-311.	1.8	100
50	POLG mutations in sporadic mitochondrial disorders with multiple mtDNA deletions. Human Mutation, 2003, 22, 498-499.	1.1	100
51	24 Month Longitudinal Data in Ambulant Boys with Duchenne Muscular Dystrophy. PLoS ONE, 2013, 8, e52512.	1.1	99
52	Embryonic stem cell-derived neural stem cells improve spinal muscular atrophy phenotype in mice. Brain, 2010, 133, 465-481.	3.7	98
53	Long Term Natural History Data in Ambulant Boys with Duchenne Muscular Dystrophy: 36-Month Changes. PLoS ONE, 2014, 9, e108205.	1.1	98
54	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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55	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
56	TARDBP (TDPâ€43) sequence analysis in patients with familial and sporadic ALS: identification of two novel mutations. European Journal of Neurology, 2009, 16, 727-732.	1.7	93
57	The genetic basis of undiagnosed muscular dystrophies and myopathies. Neurology, 2016, 87, 71-76.	1.5	92
58	Transplanted ALDHhiSSClo neural stem cells generate motor neurons and delay disease progression of nmd mice, an animal model of SMARD1. Human Molecular Genetics, 2006, 15, 167-187.	1.4	90
59	Muscle mitochondrial DNA deletion and 31P-NMR spectroscopy alterations in a migraine patient. Journal of the Neurological Sciences, 1991, 104, 182-189.	0.3	89
60	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
61	Neuroectodermal and microglial differentiation of bone marrow cells in the mouse spinal cord and sensory ganglia. Journal of Neuroscience Research, 2002, 70, 721-733.	1.3	86
62	Novel optineurin mutations in patients with familial and sporadic amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2011, 82, 1239-1243.	0.9	86
63	The italian limb girdle muscular dystrophy registry: Relative frequency, clinical features, and differential diagnosis. Muscle and Nerve, 2017, 55, 55-68.	1.0	86
64	A clinical, genetic, and biochemical characterization ofSPG7mutations in a large cohort of patients with hereditary spastic paraplegia. Human Mutation, 2008, 29, 522-531.	1.1	85
65	Mitochondrial Fusion Proteins and Human Diseases. Neurology Research International, 2013, 2013, 1-11.	0.5	85
66	A Third Locus Predisposing to Multiple Deletions of mtDNA in Autosomal Dominant Progressive External Ophthalmoplegia. American Journal of Human Genetics, 1999, 65, 256-260.	2.6	82
67	Abdominal volume contribution to tidal volume as an early indicator of respiratory impairment in Duchenne muscular dystrophy. European Respiratory Journal, 2010, 35, 1118-1125.	3.1	82
68	Growth factors in ischemic stroke. Journal of Cellular and Molecular Medicine, 2011, 15, 1645-1687.	1.6	81
69	Importance of SPP1 genotype as a covariate in clinical trials in Duchenne muscular dystrophy. Neurology, 2012, 79, 159-162.	1.5	81
70	Next-generation sequencing reveals DGUOK mutations in adult patients with mitochondrial DNA multiple deletions. Brain, 2012, 135, 3404-3415.	3.7	81
71	Mutated mitofusin 2 presents with intrafamilial variability and brain mitochondrial dysfunction. Neurology, 2008, 71, 1959-1966.	1.5	80
72	MotorPlex provides accurate variant detection across large muscle genes both in single myopathic patients and in pools of DNA samples. Acta Neuropathologica Communications, 2014, 2, 100.	2.4	76

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73	The apolipoprotein E Ïµ4 allele causes a faster decline of cognitive performances in Down's syndrome subjects. Journal of the Neurological Sciences, 1997, 145, 87-91.	0.3	75
74	Prevalence of congenital muscular dystrophy in Italy. Neurology, 2015, 84, 904-911.	1.5	75
75	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
76	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
77	Molecular Therapeutic Strategies for Spinal Muscular Atrophies: Current and Future Clinical Trials. Clinical Therapeutics, 2014, 36, 128-140.	1.1	74
78	TARDBP Mutations in Frontotemporal Lobar Degeneration: Frequency, Clinical Features, and Disease Course. Rejuvenation Research, 2010, 13, 509-517.	0.9	73
79	Modulated Generation of Neuronal Cells from Bone Marrow by Expansion and Mobilization of Circulating Stem Cells with in Vivo Cytokine Treatment. Experimental Neurology, 2002, 177, 443-452.	2.0	71
80	Human motor neuron generation from embryonic stem cells and induced pluripotent stem cells. Cellular and Molecular Life Sciences, 2010, 67, 3837-3847.	2.4	71
81	A Subpopulation of Murine Bone Marrow Cells Fully Differentiates along the Myogenic Pathway and Participates in Muscle Repair in the mdx Dystrophic Mouse. Experimental Cell Research, 2002, 277, 74-85.	1.2	70
82	Meta-analysis of vascular endothelial growth factor variations in amyotrophic lateral sclerosis: increased susceptibility in male carriers of the -2578AA genotype. Journal of Medical Genetics, 2009, 46, 840-846.	1.5	70
83	Interpreting Genetic Variants in Titin in Patients With Muscle Disorders. JAMA Neurology, 2018, 75, 557.	4.5	69
84	A missense mutation in the mitochondrial ND5 gene associated with a Leigh-MELAS overlap syndrome. Neurology, 2003, 60, 1857-1861.	1.5	68
85	Redefining phenotypes associated with mitochondrial DNA single deletion. Journal of Neurology, 2015, 262, 1301-1309.	1.8	68
86	Genome-Wide Identification of Susceptibility Alleles for Viral Infections through a Population Genetics Approach. PLoS Genetics, 2010, 6, e1000849.	1.5	67
87	Loss of Dp140 regulatory sequences is associated with cognitive impairment in dystrophinopathies. Neuromuscular Disorders, 2000, 10, 194-199.	0.3	66
88	Analysis of intronic conserved elements indicates that functional complexity might represent a major source of negative selection on non-coding sequences. Human Molecular Genetics, 2005, 14, 2533-2546.	1.4	66
89	Systemic transplantation of c-kit+ cells exerts a therapeutic effect in a model of amyotrophic lateral sclerosis. Human Molecular Genetics, 2010, 19, 3782-3796.	1.4	66
90	Mutation finding in patients with dysferlin deficiency and role of the dysferlin interacting proteins annexin A1 and A2 in muscular dystrophies. Human Mutation, 2005, 26, 283-283.	1.1	65

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91	Multipotentiality, homing properties, and pyramidal neurogenesis of CNSâ€derived LeX(sseaâ€1) + /CXCR4 + stem cells. FASEB Journal, 2005, 19, 1860-1862.	0.2	65
92	6 Minute Walk Test in Duchenne MD Patients with Different Mutations: 12 Month Changes. PLoS ONE, 2014, 9, e83400.	1.1	65
93	A New Mitochondrial DNA Mutation in ND3 Gene Causing Severe Leigh Syndrome with Early Lethality. Pediatric Research, 2004, 55, 842-846.	1.1	64
94	New Mutations in TK2 Gene Associated With Mitochondrial DNA Depletion. Pediatric Neurology, 2006, 34, 177-185.	1.0	63
95	Inclusion body myopathy and frontotemporal dementia caused by a novel VCP mutation. Neurobiology of Aging, 2009, 30, 752-758.	1.5	63
96	Coexistence of CMT-2D and distal SMA-V phenotypes in an Italian family with a GARS gene mutation. Neurology, 2006, 66, 752-754.	1.5	62
97	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
98	The signature of long-standing balancing selection at the human defensin Î²-1 promoter. Genome Biology, 2008, 9, R143.	13.9	60
99	The landscape of human genes involved in the immune response to parasitic worms. BMC Evolutionary Biology, 2010, 10, 264.	3.2	59
100	CHCHD10mutations in Italian patients with sporadic amyotrophic lateral sclerosis: Figure 1. Brain, 2015, 138, e372-e372.	3.7	59
101	Population Genetics of IFIH1: Ancient Population Structure, Local Selection, and Implications for Susceptibility to Type 1 Diabetes. Molecular Biology and Evolution, 2010, 27, 2555-2566.	3.5	58
102	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
103	Genetic Modifiers of Duchenne Muscular Dystrophy and Dilated Cardiomyopathy. PLoS ONE, 2015, 10, e0141240.	1.1	58
104	Association of a Locus in theCAMTA1Gene With Survival in Patients With Sporadic Amyotrophic Lateral Sclerosis. JAMA Neurology, 2016, 73, 812.	4.5	57
105	Metformin overdose causes platelet mitochondrial dysfunction in humans. Critical Care, 2012, 16, R180.	2.5	56
106	New motor outcome function measures in evaluation of Lateâ€Onset Pompe disease before and after enzyme replacement therapy. Muscle and Nerve, 2012, 45, 831-834.	1.0	56
107	Skeletal muscle differentiation potential of human adult bone marrow cells. Experimental Cell Research, 2004, 295, 66-78.	1.2	54
108	Is M129V of PRNP gene associated with Alzheimer's disease? A case-control study and a meta-analysis. Neurobiology of Aging, 2006, 27, 770.e1-770.e5.	1.5	54

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109	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
110	Ongoing therapeutic trials and outcome measures for Duchenne muscular dystrophy. Cellular and Molecular Life Sciences, 2013, 70, 4585-4602.	2.4	53
111	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
112	MicroRNA expression analysis identifies a subset of downregulated miRNAs in ALS motor neuron progenitors. Scientific Reports, 2018, 8, 10105.	1.6	53
113	Chemotactic Factors Enhance Myogenic Cell Migration across an Endothelial Monolayer. Experimental Cell Research, 2001, 268, 36-44.	1.2	52
114	McArdle disease: the mutation spectrum ofPYGMin a large Italian cohort. Human Mutation, 2006, 27, 718-718.	1.1	52
115	Metformin overdose, but not lactic acidosis per se, inhibits oxygen consumption in pigs. Critical Care, 2012, 16, R75.	2.5	52
116	Beta-lactam antibiotic offers neuroprotection in a spinal muscular atrophy model by multiple mechanisms. Experimental Neurology, 2011, 229, 214-225.	2.0	51
117	Both selective and neutral processes drive GC content evolution in the human genome. BMC Evolutionary Biology, 2008, 8, 99.	3.2	50
118	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
119	The Genetic Landscape of Dystrophin Mutations in Italy: A Nationwide Study. Frontiers in Genetics, 2020, 11, 131.	1.1	49
120	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
121	Intragenic Inversion of mtDNA: A New Type of Pathogenic Mutation in a Patient with Mitochondrial Myopathy. American Journal of Human Genetics, 2000, 66, 1900-1904.	2.6	48
122	Skeletal muscle gene expression profiling in mitochondrial disorders. FASEB Journal, 2005, 19, 1-30.	0.2	48
123	Molecular etiopathogenesis of limb girdle muscular and congenital muscular dystrophies: Boundaries and contiguities. Clinica Chimica Acta, 2005, 361, 54-79.	0.5	48
124	Antisense Oligonucleotide Therapy for the Treatment of C9ORF72 ALS/FTD Diseases. Molecular Neurobiology, 2014, 50, 721-732.	1.9	48
125	Fas small interfering RNA reduces motoneuron death in amyotrophic lateral sclerosis mice. Annals of Neurology, 2007, 62, 81-92.	2.8	47
126	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

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127	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
128	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
129	A third of LGMD2A biopsies have normal calpain 3 proteolytic activity as determined by an in vitro assay. Neuromuscular Disorders, 2007, 17, 148-156.	0.3	46
130	Clinical features and new molecular findings in Carnitine Palmitoyltransferase II (CPT II) deficiency. Journal of the Neurological Sciences, 2008, 266, 97-103.	0.3	46
131	Neurocognitive Profiles in Duchenne Muscular Dystrophy and Gene Mutation Site. Pediatric Neurology, 2011, 45, 292-299.	1.0	46
132	Mitochondrial Dysregulation and Impaired Autophagy in iPSC-Derived Dopaminergic Neurons of Multiple System Atrophy. Stem Cell Reports, 2018, 11, 1185-1198.	2.3	46
133	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. JAMA Neurology, 2021, 78, 1236.	4.5	46
134	A Functional Variant in ERAP1 Predisposes to Multiple Sclerosis. PLoS ONE, 2012, 7, e29931.	1.1	46
135	Retrospective study of a large population of patients affected with mitochondrial disorders: clinical, morphological and molecular genetic evaluation. Journal of Neurology, 2001, 248, 778-788.	1.8	45
136	Molecular analysis of LGMD-2B and MM patients: identification of novel DYSF mutations and possible founder effect in the Italian population. Neuromuscular Disorders, 2003, 13, 788-795.	0.3	45
137	Novel Twinkle (PEO1) gene mutations in mendelian progressive external ophthalmoplegia. Journal of Neurology, 2008, 255, 1384-1391.	1.8	45
138	Screening for later-onset Pompe's disease in patients with paucisymptomatic hyperCKemia. Molecular Genetics and Metabolism, 2013, 109, 171-173.	0.5	45
139	New genotype-phenotype correlations in a large European cohort of patients with sarcoglycanopathy. Brain, 2020, 143, 2696-2708.	3.7	45
140	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
141	TUBA4A gene analysis in sporadic amyotrophic lateral sclerosis: identification of novel mutations. Journal of Neurology, 2015, 262, 1376-1378.	1.8	44
142	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
143	Focal cognitive impairment in mitochondrial encephalomyopathies: a neuropsychological and neuroimaging study. Journal of the Neurological Sciences, 1999, 170, 57-63.	0.3	43
144	DMD Trp3X nonsense mutation associated with a founder effect in North American families with mild Becker muscular dystrophy. Neuromuscular Disorders, 2009, 19, 743-748.	0.3	43

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145	Evidence and age-related distribution of mtDNA D-loop point mutations in skeletal muscle from healthy subjects and mitochondrial patients. Journal of the Neurological Sciences, 2002, 202, 85-91.	0.3	42
146	An Evolutionary Analysis of Antigen Processing and Presentation across Different Timescales Reveals Pervasive Selection. PLoS Genetics, 2014, 10, e1004189.	1.5	42
147	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
148	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
149	Neuronal Differentiation of Murine Bone Marrow Thy-1- and Sca-1-Positive Cells. Journal of Hematotherapy and Stem Cell Research, 2003, 12, 727-734.	1.8	41
150	Absence of angiogenic genes modification in Italian ALS patients. Neurobiology of Aging, 2008, 29, 314-316.	1.5	41
151	Platelet mitochondrial dysfunction in critically ill patients: comparison between sepsis and cardiogenic shock. Critical Care, 2015, 19, 39.	2.5	41
152	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
153	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
154	Multiple deletions of mitochondrial DNA in sporadic and atypical cases of encephalomyopathy. Journal of the Neurological Sciences, 1994, 123, 74-79.	0.3	40
155	High mutational burden in the mtDNA control region from aged muscles: a single-fiber study. Neurobiology of Aging, 2003, 24, 829-838.	1.5	40
156	Intron size in mammals: complexity comes to terms with economy. Trends in Genetics, 2007, 23, 20-24.	2.9	40
157	Mitochondrial DNA G8363A mutation in the tRNALys gene: Clinical, biochemical and pathological study. Journal of the Neurological Sciences, 2009, 281, 85-92.	0.3	40
158	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.	3.1	40
159	Novel mutations at a CpG dinucleotide in the ryanodine receptor in malignant hyperthermia. , 1998, 11, 45-50.		39
160	Eight Novel Mutations in SPG4 in a Large Sample of Patients With Hereditary Spastic Paraplegia. Archives of Neurology, 2006, 63, 750.	4.9	39
161	Induced neural stem cells: Methods of reprogramming and potential therapeutic applications. Progress in Neurobiology, 2014, 114, 15-24.	2.8	39
162	Histologic muscular history in steroid-treated and untreated patients with Duchenne dystrophy. Neurology, 2015, 85, 1886-1893.	1.5	39

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163	R-Loops in Motor Neuron Diseases. Molecular Neurobiology, 2019, 56, 2579-2589.	1.9	39
164	Congenital myopathy associated with abnormal accumulation of desmin and dystrophin. Neuromuscular Disorders, 1992, 2, 169-175.	0.3	38
165	Hepatic and neuromuscular forms of glycogenosis type III: nine mutations inAGL. Human Mutation, 2006, 27, 600-601.	1.1	38
166	SPG11: a consistent clinical phenotype in a family with homozygous Spatacsin truncating mutation. Neurogenetics, 2007, 8, 301-305.	0.7	38
167	Fatigue and exercise intolerance in mitochondrial diseases. Literature revision and experience of the Italian Network of mitochondrial diseases. Neuromuscular Disorders, 2012, 22, S226-S229.	0.3	38
168	Familial idiopathic hyper-CK-emia: An underrecognized condition. Muscle and Nerve, 2006, 33, 760-765.	1.0	37
169	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
170	â€œMitochondrial neuropathiesâ€• A survey from the large cohort of the Italian Network. Neuromuscular Disorders, 2016, 26, 272-276.	0.3	37
171	Muscle glucose-6-phosphate dehydrogenase deficiency. Journal of Neurology, 1989, 236, 193-198.	1.8	36
172	Influence of the Glu298Asp polymorphism of NOS3 on age at onset and homocysteine levels in AD patients. Neurobiology of Aging, 2005, 26, 789-794.	1.5	36
173	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
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