

# Gabriele Siciliano

## List of Publications by Year in descending order

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Version: 2024-02-01

394  
papers

13,750  
citations

26630

56  
h-index

36028

97  
g-index

404  
all docs

404  
docs citations

404  
times ranked

18482  
citing authors

#	ARTICLE	IF	CITATIONS
1	A case of intravascular large B cell lymphoma with brain involvement mimicking progressive multifocal leukoencephalopathy. <i>International Journal of Neuroscience</i> , 2023, 133, 735-739.	1.6	2
2	Adult-onset mitochondrial movement disorders: a national picture from the Italian Network. <i>Journal of Neurology</i> , 2022, 269, 1413-1421.	3.6	10
3	Identification of Serum Interleukin 6 Levels as a Disease Severity Biomarker in Facioscapulohumeral Muscular Dystrophy. <i>Journal of Neuromuscular Diseases</i> , 2022, 9, 83-93.	2.6	15
4	Mutations associated with hypokalemic periodic paralysis: from hotspot regions to complete analysis of CACNA1S and SCN4A genes. <i>Neurogenetics</i> , 2022, 23, 19-25.	1.4	8
5	Validation of the DYALS (dysphagia in amyotrophic lateral sclerosis) questionnaire for the evaluation of dysphagia in ALS patients. <i>Neurological Sciences</i> , 2022, 43, 3195-3200.	1.9	3
6	Arterial intracranial thrombosis as the first manifestation of vaccine-induced immune thrombotic thrombocytopenia (VITT): a case report. <i>Neurological Sciences</i> , 2022, 43, 2085-2089.	1.9	11
7	Ictogenesis of viral pneumonia: A comparison between SARS-CoV-2 and H1N1/H3N2. <i>Epilepsy and Behavior</i> , 2022, 126, 108470.	1.7	2
8	Fatigue as a common signature of inflammatory myopathies: clinical aspects and care.. <i>Clinical and Experimental Rheumatology</i> , 2022, 40, 425-432.	0.8	4
9	Fluid Biomarkers in Alzheimer's Disease and Other Neurodegenerative Disorders: Toward Integrative Diagnostic Frameworks and Tailored Treatments. <i>Diagnostics</i> , 2022, 12, 796.	2.6	4
10	Mitochondrial Ataxias: Molecular Classification and Clinical Heterogeneity. <i>Neurology International</i> , 2022, 14, 337-356.	2.8	7
11	Association of rs3027178 polymorphism in the circadian clock gene PER1 with susceptibility to Alzheimer's disease and longevity in an Italian population. <i>GeroScience</i> , 2022, 44, 881-896.	4.6	6
12	Î±-synuclein as an emerging pathophysiological biomarker of Alzheimer's disease. <i>Expert Review of Molecular Diagnostics</i> , 2022, 22, 411-425.	3.1	4
13	Mitochondrial stroke-like episodes: The search for new therapies. <i>Pharmacological Research</i> , 2022, 180, 106228.	7.1	2
14	A Single mtDNA Deletion in Association with a LMNA Gene New Frameshift Variant: A Case Report. <i>Journal of Neuromuscular Diseases</i> , 2022, 9, 457-462.	2.6	2
15	Frailties and critical issues in neuromuscular diseases highlighted by SARS-CoV-2 pandemic: how many patients are still "invisible"?. <i>Acta Myologica</i> , 2022, 41, 24-29.	1.5	1
16	Fatigue as a common signature of inflammatory myopathies: clinical aspects and care.. <i>Clinical and Experimental Rheumatology</i> , 2022, 40, 425-432.	0.8	0
17	Is hypnotic assessment relevant to neurology?. <i>Neurological Sciences</i> , 2022, , 1.	1.9	1
18	Increase in Mitochondrial D-Loop Region Methylation Levels in Mild Cognitive Impairment Individuals. <i>International Journal of Molecular Sciences</i> , 2022, 23, 5393.	4.1	9

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19	Iron-sensitive MR imaging of the primary motor cortex to differentiate hereditary spastic paraplegia from other motor neuron diseases. <i>European Radiology</i> , 2022, 32, 8058-8064.	4.5	6
20	Therapeutic opportunities and clinical outcome measures in Duchenne muscular dystrophy. <i>Neurological Sciences</i> , 2022, 43, 625-633.	1.9	7
21	Distribution Indices of Magnetic Susceptibility Values in the Primary Motor Cortex Enable to Classify Patients with Amyotrophic Lateral Sclerosis. <i>Brain Sciences</i> , 2022, 12, 942.	2.3	4
22	Cardiac magnetic resonance in patients with muscular dystrophies. <i>European Journal of Preventive Cardiology</i> , 2021, 28, 1526-1535.	1.8	11
23	Plasma redox and inflammatory patterns during major depressive episodes: a cross-sectional investigation in elderly patients with mood disorders. <i>CNS Spectrums</i> , 2021, 26, 416-426.	1.2	3
24	Exploring the clinical association between neurological symptoms and COVID-19 pandemic outbreak: a systematic review of current literature. <i>Journal of Neurology</i> , 2021, 268, 1561-1569.	3.6	39
25	Telemedicine for neuromuscular disorders during the COVID-19 outbreak. <i>Journal of Neurology</i> , 2021, 268, 1-4.	3.6	17
26	Clinical features of mtDNA-related syndromes in adulthood. <i>Archives of Biochemistry and Biophysics</i> , 2021, 697, 108689.	3.0	10
27	Next-generation sequencing application to investigate skeletal muscle channelopathies in a large cohort of Italian patients. <i>Neuromuscular Disorders</i> , 2021, 31, 336-347.	0.6	13
28	CSF sphingomyelin: a new biomarker of demyelination in the diagnosis and management of CIDP and GBS. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2021, 92, 303-310.	1.9	20
29	Catatonia as prominent feature of stroke-like episode in MELAS. <i>Neurological Sciences</i> , 2021, 42, 383-385.	1.9	2
30	A 5-year clinical follow-up study from the Italian National Registry for FSHD. <i>Journal of Neurology</i> , 2021, 268, 356-366.	3.6	15
31	Apolipoprotein E Polymorphism and Oxidative Stress in Human Peripheral Blood Cells: Can Physical Activity Reactivate the Proteasome System through Epigenetic Mechanisms?. <i>Oxidative Medicine and Cellular Longevity</i> , 2021, 2021, 1-16.	4.0	11
32	Progress regarding the context-of-use of tau as biomarker of Alzheimer's disease and other neurodegenerative diseases. <i>Expert Review of Proteomics</i> , 2021, 18, 27-48.	3.0	8
33	Supersaturation of VEP in Migraine without Aura Patients Treated with Topiramate: An Anatomico-Functional Biomarker of the Disease. <i>Journal of Clinical Medicine</i> , 2021, 10, 769.	2.4	6
34	Obstructive sleep apnea syndrome and Alzheimer's disease pathology: may continuous positive airway pressure treatment delay cognitive deterioration?. <i>Sleep and Breathing</i> , 2021, 25, 2135-2139.	1.7	14
35	Response to levetiracetam or lamotrigine in subjects with Juvenile Myoclonic Epilepsy previously treated with valproic acid: A single center retrospective study. <i>Epilepsy and Behavior</i> , 2021, 115, 107706.	1.7	6
36	Diagnostic and prognostic value of CSF neurofilaments in a cohort of patients with motor neuron disease: A cross-sectional study. <i>Journal of Cellular and Molecular Medicine</i> , 2021, 25, 3765-3771.	3.6	10

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37	Efficacy of fingolimod after switching from interferon $\beta$ -1a in an adolescent with multiple sclerosis: case report. <i>Neurological Sciences</i> , 2021, 42, 5-7.	1.9	3
38	Autoimmune limbic encephalitis related to SARS-CoV-2 infection: Case-report and review of the literature. <i>Brain, Behavior, &amp; Immunity - Health</i> , 2021, 12, 100210.	2.5	32
39	$\beta$ -Synuclein Heteromers in Red Blood Cells of Alzheimer's Disease and Lewy Body Dementia Patients. <i>Journal of Alzheimer's Disease</i> , 2021, 80, 885-893.	2.6	9
40	Prolonged epileptic discharges predict seizure recurrence in JME: Insights from prolonged ambulatory EEG. <i>Epilepsia</i> , 2021, 62, 1184-1192.	5.1	17
41	Mitochondrial Syndromes Revisited. <i>Journal of Clinical Medicine</i> , 2021, 10, 1249.	2.4	29
42	Increased resistance towards fatigability in patients with facioscapulohumeral muscular dystrophy. <i>European Journal of Applied Physiology</i> , 2021, 121, 1617-1629.	2.5	7
43	International retrospective natural history study of <i>LMNA</i> -related congenital muscular dystrophy. <i>Brain Communications</i> , 2021, 3, fcab075.	3.3	17
44	The unfolded protein response in amyotrophic lateral sclerosis: results of a phase 2 trial. <i>Brain</i> , 2021, 144, 2635-2647.	7.6	33
45	Anti-cN1A Antibodies Are Associated with More Severe Dysphagia in Sporadic Inclusion Body Myositis. <i>Cells</i> , 2021, 10, 1146.	4.1	23
46	The neurophysiological lesson from the Italian CIDP database. <i>Neurological Sciences</i> , 2021, , 1.	1.9	3
47	SARS-CoV-2 infection in patients with primary mitochondrial diseases: Features and outcomes in Italy. <i>Mitochondrion</i> , 2021, 58, 243-245.	3.4	3
48	Movement Disorders in Children with a Mitochondrial Disease: A Cross-Sectional Survey from the Nationwide Italian Collaborative Network of Mitochondrial Diseases. <i>Journal of Clinical Medicine</i> , 2021, 10, 2063.	2.4	8
49	Cardiac magnetic resonance findings in patients with type 1 myotonic dystrophy. <i>European Heart Journal Cardiovascular Imaging</i> , 2021, 22, .	1.2	0
50	Muscle Fiber Conduction Velocity Correlates With the Age at Onset in Mild FSHD Cases. <i>Frontiers in Physiology</i> , 2021, 12, 686176.	2.8	0
51	Prolonged distal motor latency of median nerve does not improve diagnostic accuracy for CIDP. <i>Journal of Neurology</i> , 2021, , 1.	3.6	1
52	E-Health & Innovation to Overcome Barriers in Neuromuscular Diseases. Report from the 1st eNMD Congress: Nice, France, March 22-23, 2019. <i>Journal of Neuromuscular Diseases</i> , 2021, 8, 743-754.	2.6	2
53	Statins in Parkinson's Disease: Influence on Motor Progression. <i>Journal of Parkinson's Disease</i> , 2021, 11, 1651-1662.	2.8	8
54	Ultrasensitive techniques and protein misfolding amplification assays for biomarker-guided reconceptualization of Alzheimer's and other neurodegenerative diseases. <i>Expert Review of Neurotherapeutics</i> , 2021, 21, 949-967.	2.8	4

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55	Positive DAT-SCAN in SPG7: a case report mimicking possible MSA-C. <i>BMC Neurology</i> , 2021, 21, 328.	1.8	5
56	Response to “Salvage therapy for vagal nerve stimulator infection; Literature review and report of a delayed recurrence”. <i>Clinical Neurology and Neurosurgery</i> , 2021, 207, 106721.	1.4	0
57	Expanding the clinical and genetic spectrum of pathogenic variants in <i>STIM1</i> . <i>Muscle and Nerve</i> , 2021, 64, 567-575.	2.2	7
58	An updated review on the role of prescribed exercise in the management of Amyotrophic lateral sclerosis. <i>Expert Review of Neurotherapeutics</i> , 2021, 21, 871-879.	2.8	4
59	Dissecting the Interplay Between Time of Dementia and Cognitive Profiles in Lewy Body Dementias. <i>Journal of Alzheimer’s Disease</i> , 2021, 84, 757-766.	2.6	1
60	Prolonged and short epileptiform discharges have an opposite relationship with the sleep-wake cycle in patients with JME: Implications for EEG recording protocols. <i>Epilepsy and Behavior</i> , 2021, 122, 108226.	1.7	3
61	Therapeutical Management and Drug Safety in Mitochondrial Diseases” Update 2020. <i>Journal of Clinical Medicine</i> , 2021, 10, 94.	2.4	5
62	Dopamine Transporter Imaging, Current Status of a Potential Biomarker: A Comprehensive Review. <i>International Journal of Molecular Sciences</i> , 2021, 22, 11234.	4.1	19
63	CGRP Inhibitors and Oxidative Stress Biomarkers in Resistant Migraine: A Real-Life Study with Erenumab, Fremanezumab, and Galcanezumab. <i>Journal of Clinical Medicine</i> , 2021, 10, 4586.	2.4	12
64	Anti-HMGR antibodies and asymptomatic hyperCKemia. A case report. <i>Acta Myologica</i> , 2021, 40, 105-108.	1.5	1
65	Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. <i>Nature Genetics</i> , 2021, 53, 1636-1648.	21.4	223
66	31 Cardiac magnetic resonance findings in patients with Type 1 myotonic dystrophy. <i>European Heart Journal Supplements</i> , 2021, 23, .	0.1	0
67	Adapted physical activity and therapeutic exercise in late-onset Pompe disease (LOPD): a two-step rehabilitative approach. <i>Neurological Sciences</i> , 2020, 41, 859-868.	1.9	7
68	Mitochondrial epilepsy: a cross-sectional nationwide Italian survey. <i>Neurogenetics</i> , 2020, 21, 87-96.	1.4	14
69	Central Nervous System Involvement as Outcome Measure for Clinical Trials Efficacy in Myotonic Dystrophy Type 1. <i>Frontiers in Neurology</i> , 2020, 11, 624.	2.4	12
70	The role of synaptic biomarkers in the spectrum of neurodegenerative diseases. <i>Expert Review of Proteomics</i> , 2020, 17, 543-559.	3.0	16
71	Fluid Candidate Biomarkers for Alzheimer’s Disease: A Precision Medicine Approach. <i>Journal of Personalized Medicine</i> , 2020, 10, 221.	2.5	20
72	Data from the European registry for patients with McArdle disease and other muscle glycogenoses (EUROMAC). <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 330.	2.7	23

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73	Multicenter Study on Sleep and Circadian Alterations as Objective Markers of Mild Cognitive Impairment and Alzheimer's Disease Reveals Sex Differences. <i>Journal of Alzheimer's Disease</i> , 2020, 78, 1707-1719.	2.6	20
74	Different Clinical Contexts of Use of Blood Neurofilament Light Chain Protein in the Spectrum of Neurodegenerative Diseases. <i>Molecular Neurobiology</i> , 2020, 57, 4667-4691.	4.0	33
75	Impact of Coronavirus Disease 2019 Pandemic on Cognition in Parkinson's Disease. <i>Movement Disorders</i> , 2020, 35, 1717-1718.	3.9	30
76	Oxidative stress biomarkers in Fabry disease: is there a room for them?. <i>Journal of Neurology</i> , 2020, 267, 3741-3752.	3.6	12
77	Clinical and Molecular Spectrum of Myotonia and Periodic Paralysis Associated With Mutations in SCN4A in a Large Cohort of Italian Patients. <i>Frontiers in Neurology</i> , 2020, 11, 646.	2.4	7
78	Frequency of diabetes and other comorbidities in chronic inflammatory demyelinating polyradiculoneuropathy and their impact on clinical presentation and response to therapy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2020, 91, 1092-1099.	1.9	22
79	Nusinersen safety and effects on motor function in adult spinal muscular atrophy type 2 and 3. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2020, 91, 1166-1174.	1.9	99
80	Large genotype-phenotype study in carriers of D4Z4 borderline alleles provides guidance for facioscapulohumeral muscular dystrophy diagnosis. <i>Scientific Reports</i> , 2020, 10, 21648.	3.3	16
81	Primary mitochondrial myopathy. <i>Neurology: Genetics</i> , 2020, 6, e519.	1.9	10
82	Red blood cell $\alpha$ -synuclein heteroaggregates can discriminate healthy controls from cognitively impaired subjects of the AD-LBD spectrum. <i>Alzheimer's and Dementia</i> , 2020, 16, e040618.	0.8	0
83	Sex differences in red blood cell $\alpha$ -synuclein protein and its heteroaggregates with amyloid $\beta^2$ and tau in early Alzheimer's disease. <i>Alzheimer's and Dementia</i> , 2020, 16, e042079.	0.8	0
84	Assessment of the integrity of the noradrenergic nucleus locus coeruleus during normal ageing by neuromelanin- $\beta$ T MRI. <i>Alzheimer's and Dementia</i> , 2020, 16, e043332.	0.8	0
85	In vivo assessment of the noradrenergic nucleus locus coeruleus in Alzheimer's disease and other types of dementia. <i>Alzheimer's and Dementia</i> , 2020, 16, e043616.	0.8	0
86	Phenotypic Variability Among Patients With D4Z4 Reduced Allele Facioscapulohumeral Muscular Dystrophy. <i>JAMA Network Open</i> , 2020, 3, e204040.	5.9	25
87	Impact of environmental factors and physical activity on disability and quality of life in CIDP. <i>Journal of Neurology</i> , 2020, 267, 2683-2691.	3.6	4
88	Fibroblast growth factor 21 and growth differentiation factor 15 are sensitive biomarkers of mitochondrial diseases due to mitochondrial transfer-RNA mutations and mitochondrial DNA deletions. <i>Neurological Sciences</i> , 2020, 41, 3653-3662.	1.9	9
89	Exercise-Related Oxidative Stress as Mechanism to Fight Physical Dysfunction in Neuromuscular Disorders. <i>Frontiers in Physiology</i> , 2020, 11, 451.	2.8	11
90	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.	3.6	47

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91	The path to biomarker-based diagnostic criteria for the spectrum of neurodegenerative diseases. Expert Review of Molecular Diagnostics, 2020, 20, 421-441.	3.1	42
92	Relevance of diagnostic investigations in chronic inflammatory demyelinating poliradiculoneuropathy: Data from the Italian CIDP database. Journal of the Peripheral Nervous System, 2020, 25, 152-161.	3.1	15
93	Psychosocial impact of sport activity in neuromuscular disorders. Neurological Sciences, 2020, 41, 2561-2567.	1.9	8
94	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
95	Neuromuscular tetanic hyperexcitability syndrome associated to a heterozygous mutation with normal serum magnesium levels. Acta Myologica, 2020, 39, 36-39.	1.5	2
96	MRI cortical feature of bulbar impairment in patients with amyotrophic lateral sclerosis. NeuroImage: Clinical, 2019, 24, 101934.	2.7	20
97	CPEO and Mitochondrial Myopathy in a Patient with DGUOK Compound Heterozygous Pathogenetic Variant and mtDNA Multiple Deletions. Case Reports in Neurological Medicine, 2019, 2019, 1-4.	0.4	1
98	Impact of ApoE Polymorphism and Physical Activity on Plasma Antioxidant Capability and Erythrocyte Membranes. Antioxidants, 2019, 8, 538.	5.1	11
99	Plasma Levels of Oxidative Stress Markers, before and after BoNT/A Treatment, in Chronic Migraine. Toxins, 2019, 11, 608.	3.4	18
100	Phenotype may predict the clinical course of facioscapulohumeral muscular dystrophy. Muscle and Nerve, 2019, 59, 711-713.	2.2	12
101	Assessing the Role of Anti rh-GAA in Modulating Response to ERT in a Late-Onset Pompe Disease Cohort from the Italian GSDII Study Group. Advances in Therapy, 2019, 36, 1177-1189.	2.9	8
102	A novel family with axonal Charcot-Marie-Tooth disease caused by a mutation in the <i>EGR2</i> gene. Journal of the Peripheral Nervous System, 2019, 24, 219-223.	3.1	7
103	Potential Diagnostic Value of Red Blood Cells $\pm$ Synuclein Heteroaggregates in Alzheimer's Disease. Molecular Neurobiology, 2019, 56, 6451-6459.	4.0	24
104	Is early detection of late-onset Pompe disease a pneumologist's affair? A lesson from an Italian screening study. Orphanet Journal of Rare Diseases, 2019, 14, 62.	2.7	9
105	Lipomatosis Incidence and Characteristics in an Italian Cohort of Mitochondrial Patients. Frontiers in Neurology, 2019, 10, 160.	2.4	19
106	A longitudinal study of polysomnographic variables in patients with mild cognitive impairment converting to Alzheimer's disease. Journal of Sleep Research, 2019, 28, e12821.	3.2	29
107	Sleep Complaints, Sleep and Breathing Disorders in Myotonic Dystrophy Type 2. Current Neurology and Neuroscience Reports, 2019, 19, 9.	4.2	11
108	Next-generation sequencing approach to hyperCKemia. Neurology: Genetics, 2019, 5, e352.	1.9	31

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109	Pharmacogenetics of myotonic hNav1.4 sodium channel variants situated near the fast inactivation gate. <i>Pharmacological Research</i> , 2019, 141, 224-235.	7.1	25
110	Atypical CIDP: diagnostic criteria, progression and treatment response. Data from the Italian CIDP Database. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2019, 90, 125-132.	1.9	108
111	Exercise therapy in muscle diseases: open issues and future perspectives. <i>Acta Myologica</i> , 2019, 38, 233-238.	1.5	6
112	Disruption of sleep-wake continuum in myotonic dystrophy type 1: Beyond conventional sleep staging. <i>Neuromuscular Disorders</i> , 2018, 28, 414-421.	0.6	14
113	A mobile app for patients with Pompe disease and its possible clinical applications. <i>Neuromuscular Disorders</i> , 2018, 28, 471-475.	0.6	15
114	Elevated TGF $\beta$ 2 serum levels in Emery-Dreifuss Muscular Dystrophy: Implications for myocyte and tenocyte differentiation and fibrogenic processes. <i>Nucleus</i> , 2018, 9, 337-349.	2.2	25
115	$\beta$ -Synuclein Aggregates with $\beta$ -Amyloid or Tau in Human Red Blood Cells: Correlation with Antioxidant Capability and Physical Exercise in Human Healthy Subjects. <i>Molecular Neurobiology</i> , 2018, 55, 2653-2675.	4.0	32
116	Oxidative Stress Assessment in Alzheimer's Disease: A Clinic Setting Study. <i>American Journal of Alzheimer's Disease and Other Dementias</i> , 2018, 33, 35-41.	1.9	15
117	Semiautomated Evaluation of the Primary Motor Cortex in Patients with Amyotrophic Lateral Sclerosis at 3T. <i>American Journal of Neuroradiology</i> , 2018, 39, 63-69.	2.4	17
118	Proximal Myopathy due to m.5835G>A Mutation in Mitochondrial MT-TY Gene. <i>Case Reports in Neurological Medicine</i> , 2018, 2018, 1-4.	0.4	4
119	Oxidative Stress in Cerebral Small Vessel Disease Dizziness Patients, Basally and After Polyphenol Compound Supplementation. <i>Current Molecular Medicine</i> , 2018, 18, 160-165.	1.3	10
120	Functional magnetic resonance imaging with encoding task in patients with mild cognitive impairment and different severity of leukoaraiosis. <i>Psychiatry Research - Neuroimaging</i> , 2018, 282, 126-131.	1.8	5
121	$\beta$ -Synuclein Aggregated with Tau and $\beta$ -Amyloid in Human Platelets from Healthy Subjects: Correlation with Physical Exercise. <i>Frontiers in Aging Neuroscience</i> , 2018, 10, 17.	3.4	18
122	$\beta$ -Synuclein Heterocomplexes with $\beta$ -Amyloid Are Increased in Red Blood Cells of Parkinson's Disease Patients and Correlate with Disease Severity. <i>Frontiers in Molecular Neuroscience</i> , 2018, 11, 53.	2.9	51
123	Amyotrophic Lateral Sclerosis and Oxidative Stress: A Double-Blind Therapeutic Trial After Curcumin Supplementation. <i>CNS and Neurological Disorders - Drug Targets</i> , 2018, 17, 767-779.	1.4	59
124	The role of rehabilitation in the management of late-onset Pompe disease: a narrative review of the level of evidence. <i>Acta Myologica</i> , 2018, 37, 241-251.	1.5	5
125	Thyroid hormone levels in the cerebrospinal fluid correlate with disease severity in euthyroid patients with Alzheimer's disease. <i>Endocrine</i> , 2017, 55, 981-984.	2.3	21
126	Methylation analysis of DNA repair genes in Alzheimer's disease. <i>Mechanisms of Ageing and Development</i> , 2017, 161, 105-111.	4.6	16



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127	The Italian limb girdle muscular dystrophy registry: Relative frequency, clinical features, and differential diagnosis. <i>Muscle and Nerve</i> , 2017, 55, 55-68.	2.2	86
128	Novel POLG mutations and variable clinical phenotypes in 13 Italian patients. <i>Neurological Sciences</i> , 2017, 38, 563-570.	1.9	17
129	The Methylenetetrahydrofolate Reductase C677T Polymorphism and Risk for Late-Onset Alzheimer's Disease: Further Evidence in an Italian Multicenter Study. <i>Journal of Alzheimer's Disease</i> , 2017, 56, 1451-1457.	2.6	20
130	Autonomic, functional, skeletal muscle, and cardiac abnormalities are associated with increased ergoreflex sensitivity in mitochondrial disease. <i>European Journal of Heart Failure</i> , 2017, 19, 1701-1709.	7.1	18
131	Decreased Methylation of the Mitochondrial D-Loop Region in Late-Onset Alzheimer's Disease. <i>Journal of Alzheimer's Disease</i> , 2017, 59, 559-564.	2.6	66
132	Identification and characterization of three novel mutations in the <i>CASQ1</i> gene in four patients with tubular aggregate myopathy. <i>Human Mutation</i> , 2017, 38, 1761-1773.	2.5	51
133	A single center study: A $\beta$ <sup>42</sup> /p-Tau <sup>181</sup> CSF ratio to discriminate AD from FTD in clinical setting. <i>Neurological Sciences</i> , 2017, 38, 1791-1797.	1.9	16
134	Neurohormonal modulation for treatment of cardiac involvement in dystrophinopathies and mitochondrial disease. <i>European Journal of Preventive Cardiology</i> , 2017, 24, 1718-1724.	1.8	6
135	Protein misfolding, amyotrophic lateral sclerosis and guanabenz: protocol for a phase II RCT with futility design (ProMISe trial). <i>BMJ Open</i> , 2017, 7, e015434.	1.9	14
136	Amyotrophic lateral sclerosis with long lasting disease course and SOD1 and TARDBP mutations: Report of two cases and overview of the literature. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2017, 18, 137-139.	1.7	1
137	Dysphagia in Amyotrophic Lateral Sclerosis: Relationships between disease progression and Fiberoptic Endoscopic Evaluation of Swallowing. <i>Auris Nasus Larynx</i> , 2017, 44, 306-312.	1.2	30
138	Mitochondrial ANT-1 related adPEO leading to cognitive impairment: is there a link?. <i>Acta Myologica</i> , 2017, 36, 25-27.	1.5	6
139	Myotonia permanens with Nav1.4-G1306E displays varied phenotypes during course of life. <i>Acta Myologica</i> , 2017, 36, 125-134.	1.5	15
140	LOPED study: looking for an early diagnosis in a late-onset Pompe disease high-risk population. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016, 87, jnnp-2014-310164.	1.9	50
141	Gly482Ser PGC-1 $\beta$ Gene Polymorphism and Exercise-Related Oxidative Stress in Amyotrophic Lateral Sclerosis Patients. <i>Frontiers in Cellular Neuroscience</i> , 2016, 10, 102.	3.7	16
142	Clinical expression of facioscapulohumeral muscular dystrophy in carriers of 1 $\Delta$ 3 D4Z4 reduced alleles: experience of the FSHD Italian National Registry. <i>BMJ Open</i> , 2016, 6, e007798.	1.9	60
143	Biomarkers and progress of antioxidant therapy for rare mitochondrial disorders. <i>Expert Opinion on Orphan Drugs</i> , 2016, 4, 591-603.	0.8	0
144	A novel clinical tool to classify facioscapulohumeral muscular dystrophy phenotypes. <i>Journal of Neurology</i> , 2016, 263, 1204-1214.	3.6	55

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145	Mitochondrial m.3243A>G mutation and carotid artery dissection. <i>Molecular Genetics and Metabolism Reports</i> , 2016, 9, 12-14.	1.1	10
146	Mitochondrial DNA haplogroups may influence Fabry disease phenotype. <i>Neuroscience Letters</i> , 2016, 629, 58-61.	2.1	10
147	Acute encephalopathy of the temporal lobes leading to m.3243A > G. When MELAS is not always MELAS. <i>Mitochondrion</i> , 2016, 30, 148-150.	3.4	3
148	Relationship between neuropsychological impairment and grey and white matter changes in adult-onset myotonic dystrophy type 1. <i>NeuroImage: Clinical</i> , 2016, 12, 190-197.	2.7	51
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