Tanya Stojkovic

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/4336968/publications.pdf

Version: 2024-02-01

224 papers	8,643 citations	41344 49 h-index	66911 78 g-index
273 all docs	273 docs citations	273 times ranked	10084 citing authors

#	Article	IF	Citations
1	Long-term observational study of sporadic inclusion body myositis. Brain, 2011, 134, 3176-3184.	7.6	319
2	Multiple Phenotypes in Phosphoglucomutase 1 Deficiency. New England Journal of Medicine, 2014, 370, 533-542.	27.0	236
3	Mutations of the FHL1 Gene Cause Emery-Dreifuss Muscular Dystrophy. American Journal of Human Genetics, 2009, 85, 338-353.	6.2	208
4	High risk of cancer in autoimmune necrotizing myopathies: usefulness of myositis specific antibody. Brain, 2016, 139, 2131-2135.	7.6	202
5	Acute myelopathies: Clinical, laboratory and outcome profiles in 79 cases. Brain, 2001, 124, 1509-1521.	7.6	193
6	Devic's neuromyelitis optica: clinical, laboratory, MRI and outcome profile. Journal of the Neurological Sciences, 2002, 197, 57-61.	0.6	182
7	Treatment of Myasthenia Gravis Exacerbation With Intravenous Immunoglobulin. Archives of Neurology, 2005, 62, 1689.	4.5	169
8	Electrophysiological Study With Prophylactic Pacing and Survival in Adults With Myotonic Dystrophy and Conduction System Disease. JAMA - Journal of the American Medical Association, 2012, 307, 1292.	7.4	154
9	Follow-Up of Patients with History of Cervical Artery Dissection. Cerebrovascular Diseases, 1995, 5, 43-49.	1.7	148
10	Quantitative Muscle MRI as an Assessment Tool for Monitoring Disease Progression in LGMD2I: A Multicentre Longitudinal Study. PLoS ONE, 2013, 8, e70993.	2.5	148
11	A current view of the diagnosis, clinical variants, response to treatment and prognosis of chronic inflammatory demyelinating polyradiculoneuropathy. Journal of the Peripheral Nervous System, 2010, 15, 50-56.	3.1	140
12	Cerebellar ataxia, neuropathy, vestibular areflexia syndrome due to RFC1 repeat expansion. Brain, 2020, 143, 480-490.	7.6	140
13	Is Devic's neuromyelitis optica a separate disease? A comparative study with multiple sclerosis. Multiple Sclerosis Journal, 2003, 9, 521-525.	3.0	131
14	Development and Validation of a New Risk Prediction Score for Life-Threatening Ventricular Tachyarrhythmias in Laminopathies. Circulation, 2019, 140, 293-302.	1.6	131
15	COVIDâ€19â€related encephalopathy: a case series with brain FDGâ€positronâ€emission tomography/computed tomography findings. European Journal of Neurology, 2020, 27, 2651-2657.	3.3	127
16	Effect of ascorbic acid in patients with Charcotâ€“Marieâ€“Tooth disease type 1A: a multicentre, randomised, double-blind, placebo-controlled trial. Lancet Neurology, The, 2009, 8, 1103-1110.	10.2	114
17	Defects in amphiphysin 2 (BIN1) and triads in several forms of centronuclear myopathies. Acta Neuropathologica, 2011, 121, 253-266.	7.7	113
18	Early onset collagen VI myopathies: Genetic and clinical correlations. Annals of Neurology, 2010, 68, 511-520.	5.3	112

#	Article	IF	Citations
19	Patients with Familial Partial Lipodystrophy of the Dunnigan Type Due to aLMNAR482W Mutation Show Muscular and Cardiac Abnormalities. Journal of Clinical Endocrinology and Metabolism, 2004, 89, 5337-5346.	3.6	106
20	Charcot-Marie-Tooth Disease Type 2A. JAMA Neurology, 2014, 71, 1036.	9.0	105
21	Muscle Glycogenosis Due to Phosphoglucomutase 1 Deficiency. New England Journal of Medicine, 2009, 361, 425-427.	27.0	101
22	Biallelic mutations in SORD cause a common and potentially treatable hereditary neuropathy with implications for diabetes. Nature Genetics, 2020, 52, 473-481.	21.4	97
23	Intravenous corticosteroids in the postpartum period for reduction of acute exacerbations in multiple sclerosis. Multiple Sclerosis Journal, 2004, 10, 596-597.	3.0	88
24	CSF isoelectrofocusing in a large cohort of MS and other neurological diseases. European Journal of Neurology, 2004, 11, 525-529.	3.3	83
25	Electron microscopy in myofibrillar myopathies reveals clues to the mutated gene. Neuromuscular Disorders, 2008, 18, 656-666.	0.6	81
26	Muscle MRI in patients with dysferlinopathy: pattern recognition and implications for clinical trials. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 1071-1081.	1.9	81
27	Quantitative Magnetic Resonance Imaging in Limb-Girdle Muscular Dystrophy 2I: A Multinational Cross-Sectional Study. PLoS ONE, 2014, 9, e90377.	2.5	81
28	Clinical outcome in 19 French and Spanish patients with valosin-containing protein myopathy associated with Pagetâ€™s disease of bone and frontotemporal dementia. Neuromuscular Disorders, 2009, 19, 316-323.	0.6	79
29	Morphologic imaging in muscular dystrophies and inflammatory myopathies. Skeletal Radiology, 2010, 39, 1219-1227.	2.0	76
30	The Clinical Outcome Study for dysferlinopathy. Neurology: Genetics, 2016, 2, e89.	1.9	75
31	Unusual MR findings of the brain stem in arterial hypertension. American Journal of Neuroradiology, 2000, 21, 391-4.	2.4	73
32	Spastic paraplegia due to SPAST mutations is modified by the underlying mutation and sex. Brain, 2018, 141, 3331-3342.	7.6	72
33	Long-term cardiac prognosis and risk stratification in 260 adults presenting with mitochondrial diseases. European Heart Journal, 2015, 36, 2886-2893.	2.2	71
34	Guidance for the care of neuromuscular patients during the COVID-19 pandemic outbreak from the French Rare Health Care for Neuromuscular Diseases Network. Revue Neurologique, 2020, 176, 507-515.	1.5	71
35	Phenotype genotype analysis in 15 patients presenting a congenital myasthenic syndrome due to mutations in DOK7. Journal of Neurology, 2010, 257, 754-766.	3.6	70
36	Sensory chronic inflammatory demyelinating polyneuropathy: An under-recognized entity?. Muscle and Nerve, 2013, 48, 727-732.	2.2	68

#	Article	IF	Citations
37	Prospective study of patients presenting with acute partial transverse myelopathy. Journal of Neurology, 2003, 250, 1447-1452.	3.6	67
38	Multiple sclerosis and depression: influence of interferon b therapy. Multiple Sclerosis Journal, 2003, 9, 284-288.	3.0	67
39	Differential involvement of sarcomeric proteins in myofibrillar myopathies: a morphological and immunohistochemical study. Acta Neuropathologica, 2009, 117, 293-307.	7.7	67
40	Long-term follow-up of patients with congenital myasthenic syndrome caused by COLQ mutations. Neuromuscular Disorders, 2012, 22, 318-324.	0.6	64
41	SIMPLE mutation analysis in dominant demyelinating Charcot-Marie-Tooth disease: three novel mutations. Journal of the Peripheral Nervous System, 2006, 11, 148-155.	3.1	59
42	Incidence and predictors of sudden death, major conduction defects and sustained ventricular tachyarrhythmias in 1388 patients with myotonic dystrophy type 1. European Heart Journal, 2017, 38, ehw569.	2.2	59
43	Autonomic dysfunction in multiple sclerosis: cervical spinal cord atrophy correlates. Journal of Neurology, 2001, 248, 297-303.	3.6	57
44	Diaphragm: Pathophysiology and Ultrasound Imaging in Neuromuscular Disorders. Journal of Neuromuscular Diseases, 2018, 5, 1-10.	2.6	57
45	Dilated cardiomyopathy in patients with mutations in anoctamin 5. International Journal of Cardiology, 2013, 168, 76-79.	1.7	56
46	Axonal Neuropathies due to Mutations in Small Heat Shock Proteins: Clinical, Genetic, and Functional Insights into Novel Mutations. Human Mutation, 2017, 38, 556-568.	2.5	54
47	PDXK mutations cause polyneuropathy responsive to pyridoxal 5â€²â€phosphate supplementation. Annals of Neurology, 2019, 86, 225-240.	5.3	54
48	The spinal and cerebral profile of adult spinal-muscular atrophy: A multimodal imaging study. NeuroImage: Clinical, 2019, 21, 101618.	2.7	54
49	Brugada syndrome and abnormal splicing of SCN5A in myotonic dystrophy type 1. Archives of Cardiovascular Diseases, 2013, 106, 635-643.	1.6	51
50	Antibodies to clustered acetylcholine receptor: expanding the phenotype. European Journal of Neurology, 2014, 21, 130-134.	3.3	51
51	Clinical and genetic spectrum in limb-girdle muscular dystrophy type 2E. Neurology, 2015, 84, 1772-1781.	1.1	50
52	FSHD1 and FSHD2 form a disease continuum. Neurology, 2019, 92, e2273-e2285.	1.1	50
53	High cardiovascular morbidity and mortality in myofibrillar myopathies due to DES gene mutations: a 10-year longitudinal study. Neuromuscular Disorders, 2012, 22, 211-218.	0.6	49
54	Autonomic and respiratory dysfunction in Charcotâ€“Marieâ€“Tooth disease due to Thr124Met mutation in the myelin protein zero gene. Clinical Neurophysiology, 2003, 114, 1609-1614.	1.5	48

#	Article	IF	Citations
55	Heterogeneous spectrum of neuropathies in WaldenstrÃ¶m's macroglobulinemia: a diagnostic strategy to optimize their management. Journal of the Peripheral Nervous System, 2012, 17, 90-101.	3.1	47
56	MFN2-associated lipomatosis: Clinical spectrum and impact on adipose tissue. Journal of Clinical Lipidology, 2018, 12, 1420-1435.	1.5	47
57	Bi-allelic JAM2 Variants Lead to Early-Onset Recessive Primary Familial Brain Calcification. American Journal of Human Genetics, 2020, 106, 412-421.	6.2	47
58	Variants in the Oxidoreductase PYROXD1 Cause Early-Onset Myopathy with Internalized Nuclei and Myofibrillar Disorganization. American Journal of Human Genetics, 2016, 99, 1086-1105.	6.2	45
59	Autoimmune hepatitis and multiple sclerosis: a coincidental association?. Multiple Sclerosis Journal, 2005, 11, 691-693.	3.0	44
60	A novel mutation in the dynamin 2 gene in a Charcot-Marie-Tooth type 2 patient: Clinical and pathological findings. Neuromuscular Disorders, 2008, 18, 334-338.	0.6	43
61	Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. Brain, 2019, 142, 2948-2964.	7.6	43
62	European muscle MRI study in limb girdle muscular dystrophy type R1/2A (LGMDR1/LGMD2A). Journal of Neurology, 2020, 267, 45-56.	3.6	43
63	Combination of IFN?-1a (Avonexï¿½) and mycophenolate mofetil (Cellceptï¿½) in multiple sclerosis. European Journal of Neurology, 2007, 14, 85-89.	3.3	42
64	Hereditary neuropathies: An update. Revue Neurologique, 2016, 172, 775-778.	1.5	42
65	Affected female carriers of MTM1 mutations display a wide spectrum of clinical and pathological involvement: delineating diagnostic clues. Acta Neuropathologica, 2017, 134, 889-904.	7.7	42
66	Unusual ocular motor findings in multiple sclerosis. Journal of the Neurological Sciences, 2006, 243, 91-95.	0.6	41
67	Prediction of longâ€term prognosis by heteroplasmy levels of the m.3243A>G mutation in patients with the <scp>mitochondrial encephalomyopathy, lactic acidosis and strokeâ€like episodes</scp> syndrome. European Journal of Neurology, 2017, 24, 255-261.	3.3	41
68	Characteristics of clinical and electrophysiological pattern of Charcotâ€Marieâ€Tooth 4C. Journal of the Peripheral Nervous System, 2012, 17, 112-122.	3.1	40
69	Visual evoked potentials study in chronic idiopathic inflammatory demyelinating polyneuropathy. Clinical Neurophysiology, 2000, 111, 2285-2291.	1.5	39
70	Pupillary disturbances in multiple sclerosis: correlation with MRI findings. Journal of the Neurological Sciences, 2001, 188, 37-41.	0.6	39
71	Vocal cord and diaphragm paralysis, as clinical features of a French family with autosomal recessive Charot-Marie-Tooth disease, associated with a new mutation in the GDAP1 gene. Neuromuscular Disorders, 2004, 14, 261-264.	0.6	38
72	Myofibrillar myopathies: State of the art, present and future challenges. Revue Neurologique, 2015, 171, 715-729.	1.5	38

#	Article	IF	Citations
73	Muscle MRI in a large cohort of patients with oculopharyngeal muscular dystrophy. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 576-585.	1.9	38
74	Impact of Coronavirus Disease 2019 in a French Cohort of Myasthenia Gravis. Neurology, 2021, 96, e2109-e2120.	1.1	38
75	Skeletal Muscle Biopsy Analysis in Reducing Body Myopathy and Other FHL1-Related Disorders. Journal of Neuropathology and Experimental Neurology, 2013, 72, 833-845.	1.7	36
76	Natural History of Cardiac and Respiratory Involvement, Prognosis and Predictive Factors for Long-Term Survival in Adult Patients with Limb Girdle Muscular Dystrophies Type 2C and 2D. PLoS ONE, 2016, 11, e0153095.	2.5	36
77	Tubular aggregate myopathy with features of Stormorken disease due to a new STIM1 mutation. Neuromuscular Disorders, 2017, 27, 78-82.	0.6	36
78	Molecular diagnosis of inherited peripheral neuropathies by targeted next-generation sequencing: molecular spectrum delineation. BMJ Open, 2018, 8, e021632.	1.9	36
79	A multicenter retrospective study of charcotâ€marieâ€tooth disease type 4B (CMT4B) associated with mutations in myotubularinâ€related proteins (MTMRs). Annals of Neurology, 2019, 86, 55-67.	5.3	35
80	Hyperckemia and myalgia are common presentations of anoctaminâ€5â€related myopathy in French patients. Muscle and Nerve, 2017, 56, 1096-1100.	2.2	34
81	The motor unit number index (MUNIX) profile of patients with adult spinal muscular atrophy. Clinical Neurophysiology, 2018, 129, 2333-2340.	1.5	33
82	Development and Validation of a New Scoring System to Predict Survival in Patients With Myotonic Dystrophy Type 1. JAMA Neurology, 2018, 75, 573.	9.0	32
83	Sirolimus for treatment of patients with inclusion body myositis: a randomised, double-blind, placebo-controlled, proof-of-concept, phase 2b trial. Lancet Rheumatology, The, 2021, 3, e40-e48.	3.9	32
84	Motor neuron pathology in CANVAS due to RFC1 expansions. Brain, 2022, 145, 2121-2132.	7.6	32
85	One-year cyclophosphamide treatment combined with methylprednisolone improves cognitive dysfunction in progressive forms of multiple sclerosis. Multiple Sclerosis Journal, 2005, 11, 360-363.	3.0	31
86	Differentiating Emery-Dreifuss muscular dystrophy and collagen VI-related myopathies using a specific CT scanner pattern. Neuromuscular Disorders, 2010, 20, 517-523.	0.6	31
87	Expanding the importance of HMERF titinopathy: new mutations and clinical aspects. Journal of Neurology, 2019, 266, 680-690.	3.6	31
88	Specific pattern of nitric oxide synthase expression in glial cells after hippocampal injury. , 1998, 22, 329-337.		30
89	Multiple sclerosis, interferon beta and clinical thyroid dysfunction. Acta Neurologica Scandinavica, 2003, 107, 154-157.	2.1	30
90	Laminin Î±2 Deficiency-Related Muscular Dystrophy Mimicking Emery-Dreifuss andÂCollagen VI related Diseases. Journal of Neuromuscular Diseases, 2015, 2, 229-240.	2.6	30

#	Article	IF	Citations
91	GUG is an efficient initiation codon to translate the human mitochondrial ATP6 gene. Biochemical and Biophysical Research Communications, 2004, 313, 687-693.	2.1	29
92	Clinical and electrophysiological characteristics of neuropathy associated with Tangier disease. Journal of Neurology, 2012, 259, 1222-1226.	3.6	28
93	Mosaicism for Dominant Collagen 6 Mutations as a Cause for Intrafamilial Phenotypic Variability. Human Mutation, 2015, 36, 48-56.	2.5	28
94	Recessive mutations in the kinase ZAK cause a congenital myopathy with fibre type disproportion. Brain, 2017, 140, 37-48.	7.6	28
95	Natural history of limb girdle muscular dystrophy R9 over 6Âyears: searching for trial endpoints. Annals of Clinical and Translational Neurology, 2019, 6, 1033-1045.	3.7	28
96	Neutral lipid storage disease with myopathy: A whole-body nuclear MRI and metabolic study. Molecular Genetics and Metabolism, 2013, 108, 125-131.	1.1	27
97	Brain MRI in late-onset multiple sclerosis. European Journal of Neurology, 2005, 12, 241-244.	3.3	26
98	Fat and Carbohydrate Metabolism During Exercise in Phosphoglucomutase Type 1 Deficiency. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E1235-E1240.	3.6	26
99	Mutations in GFPT1-related congenital myasthenic syndromes are associated with synaptic morphological defects and underlie a tubular aggregate myopathy with synaptopathy. Journal of Neurology, 2017, 264, 1791-1803.	3.6	26
100	Diaphragm sniff ultrasound: Normal values, relationship with sniff nasal pressure and accuracy for predicting respiratory involvement in patients with neuromuscular disorders. PLoS ONE, 2019, 14, e0214288.	2.5	25
101	Upper limb onset of hereditary transthyretin amyloidosis is common in nonâ€endemic areas. European Journal of Neurology, 2019, 26, 497.	3.3	25
102	Association between prophylactic angiotensin-converting enzyme inhibitors and overall survival in Duchenne muscular dystrophyâ€"analysis of registry data. European Heart Journal, 2021, 42, 1976-1984.	2.2	25
103	Paroxysmal kinesigenic choreoathetosis as a presenting symptom of multiple sclerosis. Journal of Neurology, 2000, 247, 478-480.	3.6	24
104	Pregnancy in congenital myasthenic syndrome. Journal of Neurology, 2013, 260, 815-819.	3.6	24
105	Diagnosis of unilateral trapezius muscle palsy: 54 Cases. Muscle and Nerve, 2017, 56, 215-223.	2.2	24
106	Pathogenic Variants in the Myosin Chaperone UNC-45B Cause Progressive Myopathy with Eccentric Cores. American Journal of Human Genetics, 2020, 107, 1078-1095.	6.2	24
107	Myelopathies secondary to SjÃ¶gren's syndrome: treatment with monthly intravenous cyclophosphamide associated with corticosteroids. Journal of Rheumatology, 2006, 33, 709-11.	2.0	24
108	Guillain-BarrÃ© syndrome resembling brainstem death in a patient with brain injury. Journal of Neurology, 2001, 248, 430-432.	3.6	23

#	Article	IF	Citations
109	NIPA1 (SPG6) mutations are a rare cause of autosomal dominant spastic paraplegia in Europe. Neurogenetics, 2007, 8, 155-157.	1.4	23
110	Novel mutations in DNAJB6 cause <scp>LGMD</scp>1D and distal myopathy in French families. European Journal of Neurology, 2018, 25, 790-794.	3.3	23
111	Double-blind crossover study with dolasetron mesilate, a 5-HT 3 receptor antagonist in cerebellar syndrome secondary to multiple sclerosis. Journal of Neurology, 2003, 250, 1190-1194.	3.6	22
112	Clinical spectrum and gender differences in a large cohort of Charcotâ€“Marieâ€“Tooth type 1A patients. Journal of the Neurological Sciences, 2014, 336, 155-160.	0.6	22
113	Diagnostic power of the nonâ€ischaemic forearm exercise test in detecting glycogenosis type V. European Journal of Neurology, 2015, 22, 933-940.	3.3	22
114	Relationship between muscle impairments, postural stability, and gait parameters assessed with lower-trunk accelerometry in myotonic dystrophy type 1. Neuromuscular Disorders, 2016, 26, 428-435.	0.6	22
115	Antineoplastic agents exacerbating Charcot Marie Tooth disease: red flags to avoid permanent disability. Acta OncolÃ³gica, 2018, 57, 403-411.	1.8	22
116	Metformin rescues muscle function in BAG3 myofibrillar myopathy models. Autophagy, 2021, 17, 2494-2510.	9.1	22
117	<scp>GGPS1</scp> Mutations Cause Muscular Dystrophy/Hearing Loss/Ovarian Insufficiency Syndrome. Annals of Neurology, 2020, 88, 332-347.	5.3	22
118	Charcotâ€“Marieâ€“Tooth disease misdiagnosed as chronic inflammatory demyelinating polyradiculoneuropathy: An international multicentric retrospective study. European Journal of Neurology, 2021, 28, 2846-2854.	3.3	22
119	Effect of enzyme replacement therapy with alglucosidase alfa (MyozymeÂ®) in 12 patients with advanced late-onset Pompe disease. Molecular Genetics and Metabolism, 2017, 122, 80-85.	1.1	21
120	Genomic sequencing highlights the diverse molecular causes of Perrault syndrome: a peroxisomal disorder (PEX6), metabolic disorders (CLPP, GGPS1), and mtDNA maintenance/translation disorders (LARS2, TFAM). Human Genetics, 2020, 139, 1325-1343.	3.8	21
121	Fourth meeting of the European Neurological Society 25â€“29 June 1994 Barcelona, Spain. Journal of Neurology, 1994, 241, 1-164.	3.6	20
122	A new case of autosomal dominant myotonia associated with the V1589M missense mutation in the muscle sodium channel gene and its phenotypic classification. Neuromuscular Disorders, 2006, 16, 321-324.	0.6	20
123	Respiratory muscle dysfunction in facioscapulohumeral muscular dystrophy. Neuromuscular Disorders, 2015, 25, 632-639.	0.6	20
124	Assessment of disease progression in dysferlinopathy. Neurology, 2019, 92, .	1.1	20
125	Novel CAPN3 variant associated with an autosomal dominant calpainopathy. Neuropathology and Applied Neurobiology, 2020, 46, 564-578.	3.2	20
126	Atrial flutter in myotonic dystrophy type 1: Patient characteristics and clinical outcome. Neuromuscular Disorders, 2016, 26, 227-233.	0.6	19

#	Article	IF	Citations
127	Global versus individual muscle segmentation to assess quantitative MRI-based fat fraction changes in neuromuscular diseases. European Radiology, 2021, 31, 4264-4276.	4.5	19
128	Miyoshi myopathy and limb girdle muscular dystrophy R2 are the same disease. Neuromuscular Disorders, 2021, 31, 265-280.	0.6	18
129	Bronchiolitis obliterans with organising pneumonia during interferon Î²-1a treatment. Lancet, The, 2001, 357, 751.	13.7	17
130	Genotype and other determinants of respiratory function in myotonic dystrophy type 1. Neuromuscular Disorders, 2018, 28, 222-228.	0.6	17
131	Rigid spine syndrome associated with sensoryâ€motor axonal neuropathy resembling Charcotâ€“Marieâ€Tooth disease is characteristic of Bclâ€2â€associated athanogeneâ€3 gene mutations even without cardiac involvement. Muscle and Nerve, 2018, 57, 330-334.	2.2	17
132	Assessing Dysferlinopathy Patients Over Three Years With a New Motor Scale. Annals of Neurology, 2021, 89, 967-978.	5.3	17
133	The wide spectrum of COVID-19 neuropsychiatric complications within a multidisciplinary centre. Brain Communications, 2021, 3, fcab135.	3.3	16
134	Development of new outcome measures for adult SMA type III and IV: a multimodal longitudinal study. Journal of Neurology, 2021, 268, 1792-1802.	3.6	16
135	Cervical Spinal Cord Atrophy Profile in Adult SMN1-Linked SMA. PLoS ONE, 2016, 11, e0152439.	2.5	16
136	Impaired myocardial deformation detected by speckle-tracking echocardiography in patients with myotonic dystrophy type 1. International Journal of Cardiology, 2011, 152, 375-376.	1.7	15
137	A form of muscular dystrophy associated with pathogenic variants in JAG2. American Journal of Human Genetics, 2021, 108, 840-856.	6.2	15
138	High intra-familiar clinical variability in MORC2 mutated CMT2 patients. Brain, 2017, 140, e21-e21.	7.6	14
139	Prevalence and clinical outcomes of dystrophinâ€associated dilated cardiomyopathy without severe skeletal myopathy. European Journal of Heart Failure, 2021, 23, 1276-1286.	7.1	14
140	RFC1 repeat expansions: A recurrent cause of sensory and autonomic neuropathy with cough and ataxia. European Journal of Neurology, 2022, 29, 2156-2161.	3.3	14
141	Muscle diseases with prominent joint contractures: Main entities and diagnostic strategy. Revue Neurologique, 2013, 169, 546-563.	1.5	13
142	A novel nonsense PIEZO2 mutation in a family with scoliosis and proprioceptive defect. Neuromuscular Disorders, 2019, 29, 75-79.	0.6	13
143	Congenital myopathy with central cores and fingerprint bodies in association with malignant hyperthermia susceptibility. Neuromuscular Disorders, 2001, 11, 538-541.	0.6	12
144	Threeâ€year quantitative magnetic resonance imaging and phosphorus magnetic resonance spectroscopy study in lower limb muscle in dysferlinopathy. Journal of Cachexia, Sarcopenia and Muscle, 2022, 13, 1850-1863.	7.3	12

#	Article	IF	Citations
145	Non Random Distribution of DMD Deletion Breakpoints and Implication of Double Strand Breaks Repair and Replication Error Repair Mechanisms. Journal of Neuromuscular Diseases, 2016, 3, 227-245.	2.6	11
146	Two-dimensional electrophoresis highlights haptoglobin beta chain as an additional biomarker of congenital disorders of glycosylation. Clinica Chimica Acta, 2017, 470, 70-74.	1.1	11
147	Hearing impairment in patients with myotonic dystrophy type 2. Neurology, 2018, 90, e615-e622.	1.1	11
148	Echographic Assessment of Diaphragmatic Function in Duchenne Muscular Dystrophy from Childhood to Adulthood. Journal of Neuromuscular Diseases, 2019, 6, 55-64.	2.6	11
149	Incidence and predictors of total mortality in 267 adults presenting with mitochondrial diseases. Journal of Inherited Metabolic Disease, 2020, 43, 459-466.	3.6	11
150	Clinical correlations and longâ€term followâ€up in 100 patients with sarcoglycanopathies. European Journal of Neurology, 2021, 28, 660-669.	3.3	11
151	Congenital myopathies are mainly associated with a mild cardiac phenotype. Journal of Neurology, 2019, 266, 1367-1375.	3.6	10
152	Interferon Î²1a (AvonexÂ®) treatment in multiple sclerosis: similarity of effect on progression of disability in patients with mild and moderate disability. Journal of Neurology, 2002, 249, 184-187.	3.6	9
153	A family with a novel frameshift mutation in the PMP22 gene (c.433_434insC) causing a phenotype of hereditary neuropathy with liability to pressure palsies. Neuromuscular Disorders, 2005, 15, 493-497.	0.6	9
154	Phenotypic spectrum of Charcotâ^'Marieâ^'Tooth disease due to <scp>LITAF</scp>/<scp>SIMPLE</scp> mutations: a study of 18 patients. European Journal of Neurology, 2017, 24, 530-538.	3.3	9
155	Hereditary sensory autonomic neuropathy type II: Report of two novel mutations in the FAM134B gene. Journal of the Peripheral Nervous System, 2019, 24, 354-358.	3.1	9
156	Deep phenotyping of an international series of patients with lateâ€onset dysferlinopathy. European Journal of Neurology, 2021, 28, 2092-2102.	3.3	9
157	Charcot-Marie-Tooth disease type 2CC due toNEFHvariants causes a progressive, non-length-dependent, motor-predominant phenotype. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, 48-56.	1.9	9
158	High-Throughput Digital Image Analysis Reveals Distinct Patterns of Dystrophin Expression in Dystrophinopathy Patients. Journal of Neuropathology and Experimental Neurology, 2021, 80, 955-965.	1.7	9
159	A multicenter cross-sectional French study of the impact of COVID-19 on neuromuscular diseases. Orphanet Journal of Rare Diseases, 2021, 16, 450.	2.7	9
160	Severe and rapidly evolving peripheral neuropathy revealing sporadic Creutzfeldt-Jakob disease. Journal of Neurology, 2009, 256, 134-136.	3.6	8
161	A new congenital multicore titinopathy associated with fast myosin heavy chain deficiency. Annals of Clinical and Translational Neurology, 2020, 7, 846-854.	3.7	8
162	Periorbital Ecchymoses Are Not Pathognomonic of the Light-chain Type of Amyloidosis. Acta Dermato-Venereologica, 2007, 87, 544-545.	1.3	7

#	Article	IF	Citations
163	Peripheral neuropathy in glycogen storage disease type III: Fact or myth?. Muscle and Nerve, 2016, 53, 310-312.	2.2	7
164	Dilated cardiomyopathy and limb-girdle muscular dystrophy-dystroglycanopathy due to novel pathogenic variants in the DPM3 gene. Neuromuscular Disorders, 2019, 29, 497-502.	0.6	7
165	A new case of SMA phenotype without epilepsy due to biallelic variants in ASAH1. European Journal of Human Genetics, 2019, 27, 337-339.	2.8	7
166	Genetic Characterization of a French Cohort of GNEâ€mutation negative inclusion body myopathy patients with exome sequencing. Muscle and Nerve, 2017, 56, 993-997.	2.2	6
167	Novel Phenotypes and Cardiac Involvement Associated With DNA2 Genetic Variants. Frontiers in Neurology, 2019, 10, 1049.	2.4	6
168	Missense mutations in small muscle protein X-linked (SMPX) cause distal myopathy with protein inclusions. Acta Neuropathologica, 2021, 142, 375-393.	7.7	6
169	Genotypeâ€“phenotype correlation in French patients with myelin protein zero geneâ€related inherited neuropathy. European Journal of Neurology, 2021, 28, 2913-2921.	3.3	6
170	Clinical and Molecular Spectrum Associated with COL6A3 c.7447A>G p.(Lys2483Glu) Variant: Elucidating its Role in Collagen VI-related Myopathies. Journal of Neuromuscular Diseases, 2021, 8, 633-645.	2.6	6
171	Risk for Complications after Pacemaker or Cardioverter Defibrillator Implantations in Patients with Myotonic Dystrophy Type 1. Journal of Neuromuscular Diseases, 2017, 4, 175-181.	2.6	5
172	WES homozygosity mapping in a recessive form of Charcot-Marie-Tooth neuropathy reveals intronic GDAP1 variant leading to a premature stop codon. Neurogenetics, 2018, 19, 67-76.	1.4	5
173	The role of electrodiagnosis with long exercise test in mcardle disease. Muscle and Nerve, 2018, 58, 64-71.	2.2	5
174	LRSAM1 variants and founder effect in French families with ataxic form of Charcot-Marie-Tooth type 2. European Journal of Human Genetics, 2019, 27, 1406-1418.	2.8	5
175	A high prevalence of arterial hypertension in patients with mitochondrial diseases. Journal of Inherited Metabolic Disease, 2020, 43, 478-485.	3.6	5
176	Mutation m.3395Aâ€‰>â€‰G in MT-ND1 leads to variable pathologic manifestations. Human Molecular Genetics, 2020, 29, 980-989.	2.9	5
177	Diagnostic interest of whole-body MRI in early- and late-onset LAMA2 muscular dystrophies: a large international cohort. Journal of Neurology, 2022, 269, 2414-2429.	3.6	5
178	Unravelling the impact of frontal lobe impairment for social dysfunction in myotonic dystrophy type 1. Brain Communications, 2022, 4, .	3.3	5
179	The diagnostic value of hyperammonaemia induced by the non-ischaemic forearm exercise test. Journal of Clinical Pathology, 2017, 70, 896-898.	2.0	4
180	Ganglionopathies Associated with MERRF Syndrome: An Original Report. Journal of Neuromuscular Diseases, 2020, 7, 419-423.	2.6	4

#	Article	IF	Citations
181	Assessment of diaphragm motion using ultrasonography in a patient with facio-scapulo-humeral dystrophy. Medicine (United States), 2019, 98, e13887.	1.0	3
182	A novel PHKA1 mutation associating myopathy and cognitive impairment: Expanding the spectrum of phosphorylase kinase b (PhK) deficiency. Journal of the Neurological Sciences, 2021, 424, 117391.	0.6	3
183	Improved Cardiac Outcomes by Early Treatment with Angiotensin-Converting Enzyme Inhibitors in Becker Muscular Dystrophy. Journal of Neuromuscular Diseases, 2021, 8, 495-502.	2.6	3
184	Leukoencephalopathy and conduction blocks in PLEKHG5-associated intermediate CMT disease. Neuromuscular Disorders, 2021, 31, 756-764.	0.6	3
185	Determinants of diaphragm inspiratory motion, diaphragm thickening, and its performance for predicting respiratory restrictive pattern in <scp>Duchenne</scp> muscular dystrophy. Muscle and Nerve, 2022, 65, 89-95.	2.2	3
186	Intensive Teenage Activity Is Associated With Greater Muscle Hyperintensity on T1W Magnetic Resonance Imaging in Adults With Dysferlinopathy. Frontiers in Neurology, 2020, 11, 613446.	2.4	3
187	Ullrich Congenital Dystrophy and Bethlem Myopathy: Current Knowledge on the Clinical Spectrum, Pathogenesis, and Future Therapeutic Avenues of Collagen VI Related Muscular Dystrophies. Current Pediatric Reviews, 2009, 5, 28-35.	0.8	2
188	Isokinetic assessment of trunk muscles in facioscapulohumeral muscular dystrophy type 1 patients. Neuromuscular Disorders, 2018, 28, 996-1002.	0.6	2
189	High Risk of Fatal and Nonfatal Venous Thromboembolism in Myotonic Dystrophy. Circulation, 2018, 138, 1169-1171.	1.6	2
190	Demyelinating Charcotâ€“Marieâ€“Tooth neuropathy associated with FBLN5 mutations. European Journal of Neurology, 2020, 27, 2568-2574.	3.3	2
191	Brody myopathy demonstrates a pseudoâ€increment on repetitive nerve stimulation. Muscle and Nerve, 2020, 61, 491-495.	2.2	2
192	Cramp-fasciculation syndrome phenotype of cerebellar ataxia with neuropathy and vestibular areflexia syndrome (CANVAS) due to RFC1 repeat expansion. Clinical Neurophysiology, 2022, 134, 34-36.	1.5	2
193	Multiple sclerosis with normalneuro-ophthalmological work-up:results of focal stimulations inducedby a scanning laser ophthalmoscope. Acta Neurologica Scandinavica, 2000, 101, 311-314.	2.1	1
194	M.P.4.02 Permanent muscle weakness in McArdle disease. Neuromuscular Disorders, 2007, 17, 859-860.	0.6	1
195	P5.17 Congenital Myasthenic Syndromes with COLQ mutations: Long term follow-up. Neuromuscular Disorders, 2011, 21, 728-729.	0.6	1
196	Anoctamin 5 myopathy: More patients, more phenotypes. Journal of the Neurological Sciences, 2013, 333, e447.	0.6	1
197	A novel mutation in DNAJB6 causes LGMD1D in two French families. Neuromuscular Disorders, 2015, 25, S236.	0.6	1
198	TRPV4 gene polymorphism as a phenotype modifier in a family with COL6-linked Bethlem myopathy. Neuromuscular Disorders, 2016, 26, S188.	0.6	1

#	Article	IF	Citations
199	A novel DNAJB6 mutation causing variable phenotypic expression: From distal myopathy to limb girdle muscular dystrophy. Neuromuscular Disorders, 2016, 26, S93-S94.	0.6	1
200	Motor neuron disease of very long disease duration or Charcotâ€“Marieâ€“Tooth disease? A novel phenotype related to the SOD1 p.E22G variant. Revue Neurologique, 2017, 173, 671-673.	1.5	1
201	Is cardiac dysfunction a feature of dysferlinopathy? Data from the Clinical Outcome Study of Dysferlinopathy. Neuromuscular Disorders, 2018, 28, S5-S6.	0.6	1
202	Scapular dyskinesis in myotonic dystrophy type 1: clinical characteristics and genetic investigations. Journal of Neurology, 2019, 266, 2987-2996.	3.6	1
203	Confounding clinical presentation and different disease progression in CMT4B1. Neuromuscular Disorders, 2020, 30, 576-582.	0.6	1
204	Phenotypical variability and atypical presentations in a French cohort of Andersenâ€“Tawil syndrome. European Journal of Neurology, 2022, 29, 2398-2411.	3.3	1
205	M.P.2.13 The French Pompe registry: A prospective clinical survey of patients with late onset Pompe disease in France. Neuromuscular Disorders, 2007, 17, 796-797.	0.6	0
206	EM.P.4.02 Comprehensive clinical, cellular and molecular assessment of 64 French families with COL6-related muscle disorders: Clues for genotype/phenotype correlations. Neuromuscular Disorders, 2009, 19, 606-607.	0.6	0
207	G.P.16.06 Anti-SRP antibody myopathies: clinical characteristics and follow-up of 24 patients. Neuromuscular Disorders, 2009, 19, 652-653.	0.6	0
208	P2.37 Long term prognosis of congenital myasthenic syndromes (CMS): The French CMS network experience. Neuromuscular Disorders, 2010, 20, 629-630.	0.6	0
209	Erratum to â€˜Clinical outcome in 19 French and Spanish patients with valosin-containing protein myopathy associated with Pagetâ€™s disease of bone and frontotemporal dementiaâ€™ [Neuromuscular Disorders 19 (2009) 316â€“323]. Neuromuscular Disorders, 2011, 21, e1.	0.6	0
210	P.12.6 Congenital myasthenic syndromes: Diagnosis difficulties, course and prognosis, and therapy â€“ The French CMS network experience. Neuromuscular Disorders, 2013, 23, 806-807.	0.6	0
211	G.P.72. Neuromuscular Disorders, 2014, 24, 816.	0.6	0
212	G.P.268. Neuromuscular Disorders, 2014, 24, 897.	0.6	0
213	Charcotâ€“Marieâ€“Tooth type 4B1 (MTMR2 gene): Confounding clinical presentation and report of 5 original mutations. Neuromuscular Disorders, 2015, 25, S285.	0.6	0
214	Reversible endogenous downregulation of myostatin pathway in wasting neuromuscular diseases explains challenges of anti-myostatin therapeutic approaches. Neuromuscular Disorders, 2017, 27, S97-S98.	0.6	0
215	Corrigendum to â€œ22nd International Congress of the World Muscle Society, Saint Malo, France, 3rdâ€“7th October 2017â€•[Neuromuscular Disorders 27S2 (2017) S51â€“S270]. Neuromuscular Disorders, 2017, 27, e1.	0.6	0
216	Longitudinal upper limb muscle MRI in dysferlinopathy: examining the relationship between semi quantitative MRI and physiotherapy outcome measures. Neuromuscular Disorders, 2018, 28, S6-S7.	0.6	0

#	Article	IF	Citations
217	Non invasive mechanical ventilation in DM1: The strong correlation between lung function, neurological-cognitive function and CTG repeats. Neuromuscular Disorders, 2018, 28, 894-895.	0.6	0
218	MRI â€“ MUSCLE IMAGING. Neuromuscular Disorders, 2019, 29, S153-S154.	0.6	0
219	DISORDERS OF THE EXTRACELLULAR MATRIX. Neuromuscular Disorders, 2019, 29, S192-S193.	0.6	0
220	SMA: REGISTRIES, BIOMARKERS & OUTCOME MEASURES. Neuromuscular Disorders, 2020, 30, S99-S100.	0.6	0
221	CONGENITAL MUSCULAR DYSTROPHIES. Neuromuscular Disorders, 2020, 30, S105-S106.	0.6	0
222	NEW GENES AND DISEASES. Neuromuscular Disorders, 2021, 31, S141-S142.	0.6	0
223	OTHER NMDs. Neuromuscular Disorders, 2021, 31, S156.	0.6	0
224	CHANNELOPATHIES AND RELATED DISORDERS. Neuromuscular Disorders, 2021, 31, S116.	0.6	0