# Tanya Stojkovic

## List of Publications by Year in descending order

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Version: 2024-02-01

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|---|---|---|---|
| 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 | 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 |
| 2 | 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 |
| 3 | 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 |
| 4 | 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 |
| 5 | 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 |
| 6 | 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 |
| 7 | Motor neuron pathology in CANVAS due to RFC1 expansions. Brain, 2022, 145, 2121-2132. | 7.6 | 32 |
| 8 | Phenotypical variability and atypical presentations in a French cohort of Andersenâ€“Tawil syndrome. European Journal of Neurology, 2022, 29, 2398-2411. | 3.3 | 1 |
| 9 | Unravelling the impact of frontal lobe impairment for social dysfunction in myotonic dystrophy type 1. Brain Communications, 2022, 4, . | 3.3 | 5 |
| 10 | Metformin rescues muscle function in BAG3 myofibrillar myopathy models. Autophagy, 2021, 17, 2494-2510. | 9.1 | 22 |
| 11 | Clinical correlations and longâ€term followâ€up in 100 patients with sarcoglycanopathies. European Journal of Neurology, 2021, 28, 660-669. | 3.3 | 11 |
| 12 | 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 |
| 13 | 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 |
| 14 | The wide spectrum of COVID-19 neuropsychiatric complications within a multidisciplinary centre. Brain Communications, 2021, 3, fcab135. | 3.3 | 16 |
| 15 | 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 |
| 16 | Impact of Coronavirus Disease 2019 in a French Cohort of Myasthenia Gravis. Neurology, 2021, 96, e2109-e2120. | 1.1 | 38 |
| 17 | Assessing Dysferlinopathy Patients Over Three Years With a New Motor Scale. Annals of Neurology, 2021, 89, 967-978. | 5.3 | 17 |
| 18 | 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Deep phenotyping of an international series of patients with lateâ€onset dysferlinopathy. European Journal of Neurology, 2021, 28, 2092-2102. | 3.3 | 9 |
| 20 | Miyoshi myopathy and limb girdle muscular dystrophy R2 are the same disease. Neuromuscular Disorders, 2021, 31, 265-280. | 0.6 | 18 |
| 21 | 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 |
| 22 | Missense mutations in small muscle protein X-linked (SMPX) cause distal myopathy with protein inclusions. Acta Neuropathologica, 2021, 142, 375-393. | 7.7 | 6 |
| 23 | A form of muscular dystrophy associated with pathogenic variants in JAG2. American Journal of Human Genetics, 2021, 108, 840-856. | 6.2 | 15 |
| 24 | 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 |
| 25 | 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 |
| 26 | 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 |
| 27 | 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 |
| 28 | 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 |
| 29 | Leukoencephalopathy and conduction blocks in PLEKHG5-associated intermediate CMT disease. Neuromuscular Disorders, 2021, 31, 756-764. | 0.6 | 3 |
| 30 | 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 |
| 31 | NEW GENES AND DISEASES. Neuromuscular Disorders, 2021, 31, S141-S142. | 0.6 | 0 |
| 32 | OTHER NMDs. Neuromuscular Disorders, 2021, 31, S156. | 0.6 | 0 |
| 33 | CHANNELOPATHIES AND RELATED DISORDERS. Neuromuscular Disorders, 2021, 31, S116. | 0.6 | 0 |
| 34 | 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 |
| 35 | European muscle MRI study in limb girdle muscular dystrophy type R1/2A (LGMDR1/LGMD2A). Journal of Neurology, 2020, 267, 45-56. | 3.6 | 43 |
| 36 | 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 |

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|---|---|---|---|
| 37 | A high prevalence of arterial hypertension in patients with mitochondrial diseases. Journal of Inherited Metabolic Disease, 2020, 43, 478-485. | 3.6 | 5 |
| 38 | SMA: REGISTRIES, BIOMARKERS & OUTCOME MEASURES. Neuromuscular Disorders, 2020, 30, S99-S100. | 0.6 | 0 |
| 39 | CONGENITAL MUSCULAR DYSTROPHIES. Neuromuscular Disorders, 2020, 30, S105-S106. | 0.6 | 0 |
| 40 | 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 |
| 41 | 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 |
| 42 | Demyelinating Charcotâ€“Marieâ€“Tooth neuropathy associated with FBLN5 mutations. European Journal of Neurology, 2020, 27, 2568-2574. | 3.3 | 2 |
| 43 | 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 |
| 44 | <scp>GGPS1</scp> Mutations Cause Muscular Dystrophy/Hearing Loss/Ovarian Insufficiency Syndrome. Annals of Neurology, 2020, 88, 332-347. | 5.3 | 22 |
| 45 | 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 |
| 46 | Ganglionopathies Associated with MERRF Syndrome: An Original Report. Journal of Neuromuscular Diseases, 2020, 7, 419-423. | 2.6 | 4 |
| 47 | 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 |
| 48 | Confounding clinical presentation and different disease progression in CMT4B1. Neuromuscular Disorders, 2020, 30, 576-582. | 0.6 | 1 |
| 49 | Mutation m.3395Aâ€‰>â€‰G in MT-ND1 leads to variable pathologic manifestations. Human Molecular Genetics, 2020, 29, 980-989. | 2.9 | 5 |
| 50 | Brody myopathy demonstrates a pseudoâ€increment on repetitive nerve stimulation. Muscle and Nerve, 2020, 61, 491-495. | 2.2 | 2 |
| 51 | Cerebellar ataxia, neuropathy, vestibular areflexia syndrome due to RFC1 repeat expansion. Brain, 2020, 143, 480-490. | 7.6 | 140 |
| 52 | Novel CAPN3 variant associated with an autosomal dominant calpainopathy. Neuropathology and Applied Neurobiology, 2020, 46, 564-578. | 3.2 | 20 |
| 53 | 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 |
| 54 | 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 |

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|---|---|---|---|
| 55 | 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 |
| 56 | MRI â€“ MUSCLE IMAGING. Neuromuscular Disorders, 2019, 29, S153-S154. | 0.6 | 0 |
| 57 | DISORDERS OF THE EXTRACELLULAR MATRIX. Neuromuscular Disorders, 2019, 29, S192-S193. | 0.6 | 0 |
| 58 | 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 |
| 59 | Novel Phenotypes and Cardiac Involvement Associated With DNA2 Genetic Variants. Frontiers in Neurology, 2019, 10, 1049. | 2.4 | 6 |
| 60 | Scapular dyskinesis in myotonic dystrophy type 1: clinical characteristics and genetic investigations. Journal of Neurology, 2019, 266, 2987-2996. | 3.6 | 1 |
| 61 | Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. Brain, 2019, 142, 2948-2964. | 7.6 | 43 |
| 62 | Expanding the importance of HMERF titinopathy: new mutations and clinical aspects. Journal of Neurology, 2019, 266, 680-690. | 3.6 | 31 |
| 63 | Development and Validation of a New Risk Prediction Score for Life-Threatening Ventricular Tachyarrhythmias in Laminopathies. Circulation, 2019, 140, 293-302. | 1.6 | 131 |
| 64 | PDXK mutations cause polyneuropathy responsive to pyridoxal 5â€²â€phosphate supplementation. Annals of Neurology, 2019, 86, 225-240. | 5.3 | 54 |
| 65 | 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 |
| 66 | 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 |
| 67 | 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 |
| 68 | 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 |
| 69 | FSHD1 and FSHD2 form a disease continuum. Neurology, 2019, 92, e2273-e2285. | 1.1 | 50 |
| 70 | 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 |
| 71 | Congenital myopathies are mainly associated with a mild cardiac phenotype. Journal of Neurology, 2019, 266, 1367-1375. | 3.6 | 10 |
| 72 | Assessment of diaphragm motion using ultrasonography in a patient with facio-scapulo-humeral dystrophy. Medicine (United States), 2019, 98, e13887. | 1.0 | 3 |

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|---|---|---|---|
| 73 | Echographic Assessment of Diaphragmatic Function in Duchenne Muscular Dystrophy from Childhood to Adulthood. Journal of Neuromuscular Diseases, 2019, 6, 55-64. | 2.6 | 11 |
| 74 | A novel nonsense PIEZO2 mutation in a family with scoliosis and proprioceptive defect. Neuromuscular Disorders, 2019, 29, 75-79. | 0.6 | 13 |
| 75 | Assessment of disease progression in dysferlinopathy. Neurology, 2019, 92, . | 1.1 | 20 |
| 76 | Upper limb onset of hereditary transthyretin amyloidosis is common in nonâ€endemic areas. European Journal of Neurology, 2019, 26, 497. | 3.3 | 25 |
| 77 | 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 |
| 78 | The spinal and cerebral profile of adult spinal-muscular atrophy: A multimodal imaging study. NeuroImage: Clinical, 2019, 21, 101618. | 2.7 | 54 |
| 79 | 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 |
| 80 | 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 |
| 81 | 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 |
| 82 | 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 |
| 83 | Hearing impairment in patients with myotonic dystrophy type 2. Neurology, 2018, 90, e615-e622. | 1.1 | 11 |
| 84 | The role of electrodiagnosis with long exercise test in mcardle disease. Muscle and Nerve, 2018, 58, 64-71. | 2.2 | 5 |
| 85 | Genotype and other determinants of respiratory function in myotonic dystrophy type 1. Neuromuscular Disorders, 2018, 28, 222-228. | 0.6 | 17 |
| 86 | Diaphragm: Pathophysiology and Ultrasound Imaging in Neuromuscular Disorders. Journal of Neuromuscular Diseases, 2018, 5, 1-10. | 2.6 | 57 |
| 87 | Antineoplastic agents exacerbating Charcot Marie Tooth disease: red flags to avoid permanent disability. Acta OncolÃ³gica, 2018, 57, 403-411. | 1.8 | 22 |
| 88 | 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 |
| 89 | Spastic paraplegia due to SPAST mutations is modified by the underlying mutation and sex. Brain, 2018, 141, 3331-3342. | 7.6 | 72 |
| 90 | Isokinetic assessment of trunk muscles in facioscapulohumeral muscular dystrophy type 1 patients. Neuromuscular Disorders, 2018, 28, 996-1002. | 0.6 | 2 |

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|---|---|---|---|
| 91 | High Risk of Fatal and Nonfatal Venous Thromboembolism in Myotonic Dystrophy. Circulation, 2018, 138, 1169-1171. | 1.6 | 2 |
| 92 | Molecular diagnosis of inherited peripheral neuropathies by targeted next-generation sequencing: molecular spectrum delineation. BMJ Open, 2018, 8, e021632. | 1.9 | 36 |
| 93 | The motor unit number index (MUNIX) profile of patients with adult spinal muscular atrophy. Clinical Neurophysiology, 2018, 129, 2333-2340. | 1.5 | 33 |
| 94 | MFN2-associated lipomatosis: Clinical spectrum and impact on adipose tissue. Journal of Clinical Lipidology, 2018, 12, 1420-1435. | 1.5 | 47 |
| 95 | 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 |
| 96 | Is cardiac dysfunction a feature of dysferlinopathy? Data from the Clinical Outcome Study of Dysferlinopathy. Neuromuscular Disorders, 2018, 28, S5-S6. | 0.6 | 1 |
| 97 | 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 |
| 98 | 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 |
| 99 | 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 |
| 100 | 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 |
| 101 | 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 |
| 102 | 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 |
| 103 | Hyperckemia and myalgia are common presentations of anoctaminâ€5â€related myopathy in French patients. Muscle and Nerve, 2017, 56, 1096-1100. | 2.2 | 34 |
| 104 | The diagnostic value of hyperammonaemia induced by the non-ischaemic forearm exercise test. Journal of Clinical Pathology, 2017, 70, 896-898. | 2.0 | 4 |
| 105 | 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 |
| 106 | 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 |
| 107 | High intra-familiar clinical variability in MORC2 mutated CMT2 patients. Brain, 2017, 140, e21-e21. | 7.6 | 14 |
| 108 | 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 |

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|---|---|---|---|
| 109 | 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 |
| 110 | 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 |
| 111 | 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 |
| 112 | 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 |
| 113 | Diagnosis of unilateral trapezius muscle palsy: 54 Cases. Muscle and Nerve, 2017, 56, 215-223. | 2.2 | 24 |
| 114 | Recessive mutations in the kinase ZAK cause a congenital myopathy with fibre type disproportion. Brain, 2017, 140, 37-48. | 7.6 | 28 |
| 115 | Tubular aggregate myopathy with features of Stormorken disease due to a new STIM1 mutation. Neuromuscular Disorders, 2017, 27, 78-82. | 0.6 | 36 |
| 116 | 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 |
| 117 | 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 |
| 118 | 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 |
| 119 | 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 |
| 120 | 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 |
| 121 | High risk of cancer in autoimmune necrotizing myopathies: usefulness of myositis specific antibody. Brain, 2016, 139, 2131-2135. | 7.6 | 202 |
| 122 | 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 |
| 123 | Hereditary neuropathies: An update. Revue Neurologique, 2016, 172, 775-778. | 1.5 | 42 |
| 124 | The Clinical Outcome Study for dysferlinopathy. Neurology: Genetics, 2016, 2, e89. | 1.9 | 75 |
| 125 | TRPV4 gene polymorphism as a phenotype modifier in a family with COL6-linked Bethlem myopathy. Neuromuscular Disorders, 2016, 26, S188. | 0.6 | 1 |
| 126 | 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 |

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|---|---|---|---|
| 127 | Peripheral neuropathy in glycogen storage disease type III: Fact or myth?. Muscle and Nerve, 2016, 53, 310-312. | 2.2 | 7 |
| 128 | Atrial flutter in myotonic dystrophy type 1: Patient characteristics and clinical outcome. Neuromuscular Disorders, 2016, 26, 227-233. | 0.6 | 19 |
| 129 | Cervical Spinal Cord Atrophy Profile in Adult SMN1-Linked SMA. PLoS ONE, 2016, 11, e0152439. | 2.5 | 16 |
| 130 | Mosaicism for Dominant Collagen 6 Mutations as a Cause for Intrafamilial Phenotypic Variability. Human Mutation, 2015, 36, 48-56. | 2.5 | 28 |
| 131 | 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 |
| 132 | Respiratory muscle dysfunction in facioscapulohumeral muscular dystrophy. Neuromuscular Disorders, 2015, 25, 632-639. | 0.6 | 20 |
| 133 | Long-term cardiac prognosis and risk stratification in 260 adults presenting with mitochondrial diseases. European Heart Journal, 2015, 36, 2886-2893. | 2.2 | 71 |
| 134 | Clinical and genetic spectrum in limb-girdle muscular dystrophy type 2E. Neurology, 2015, 84, 1772-1781. | 1.1 | 50 |
| 135 | Myofibrillar myopathies: State of the art, present and future challenges. Revue Neurologique, 2015, 171, 715-729. | 1.5 | 38 |
| 136 | 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 |
| 137 | A novel mutation in DNAJB6 causes LGMD1D in two French families. Neuromuscular Disorders, 2015, 25, S236. | 0.6 | 1 |
| 138 | Charcotâ€“Marieâ€“Tooth type 4B1 (MTMR2 gene): Confounding clinical presentation and report of 5 original mutations. Neuromuscular Disorders, 2015, 25, S285. | 0.6 | 0 |
| 139 | Antibodies to clustered acetylcholine receptor: expanding the phenotype. European Journal of Neurology, 2014, 21, 130-134. | 3.3 | 51 |
| 140 | Charcot-Marie-Tooth Disease Type 2A. JAMA Neurology, 2014, 71, 1036. | 9.0 | 105 |
| 141 | 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 |
| 142 | Multiple Phenotypes in Phosphoglucomutase 1 Deficiency. New England Journal of Medicine, 2014, 370, 533-542. | 27.0 | 236 |
| 143 | G.P.72. Neuromuscular Disorders, 2014, 24, 816. | 0.6 | 0 |
| 144 | G.P.268. Neuromuscular Disorders, 2014, 24, 897. | 0.6 | 0 |

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|---|---|---|---|
| 145 | Quantitative Magnetic Resonance Imaging in Limb-Girdle Muscular Dystrophy 2I: A Multinational Cross-Sectional Study. PLoS ONE, 2014, 9, e90377. | 2.5 | 81 |
| 146 | Pregnancy in congenital myasthenic syndrome. Journal of Neurology, 2013, 260, 815-819. | 3.6 | 24 |
| 147 | Brugada syndrome and abnormal splicing of SCN5A in myotonic dystrophy type 1. Archives of Cardiovascular Diseases, 2013, 106, 635-643. | 1.6 | 51 |
| 148 | Muscle diseases with prominent joint contractures: Main entities and diagnostic strategy. Revue Neurologique, 2013, 169, 546-563. | 1.5 | 13 |
| 149 | Sensory chronic inflammatory demyelinating polyneuropathy: An under-recognized entity?. Muscle and Nerve, 2013, 48, 727-732. | 2.2 | 68 |
| 150 | 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 |
| 151 | Dilated cardiomyopathy in patients with mutations in anoctamin 5. International Journal of Cardiology, 2013, 168, 76-79. | 1.7 | 56 |
| 152 | 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 |
| 153 | Anoctamin 5 myopathy: More patients, more phenotypes. Journal of the Neurological Sciences, 2013, 333, e447. | 0.6 | 1 |
| 154 | Fat and Carbohydrate Metabolism During Exercise in Phosphoglucomutase Type 1 Deficiency. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E1235-E1240. | 3.6 | 26 |
| 155 | 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 |
| 156 | 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 |
| 157 | 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 |
| 158 | Clinical and electrophysiological characteristics of neuropathy associated with Tangier disease. Journal of Neurology, 2012, 259, 1222-1226. | 3.6 | 28 |
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