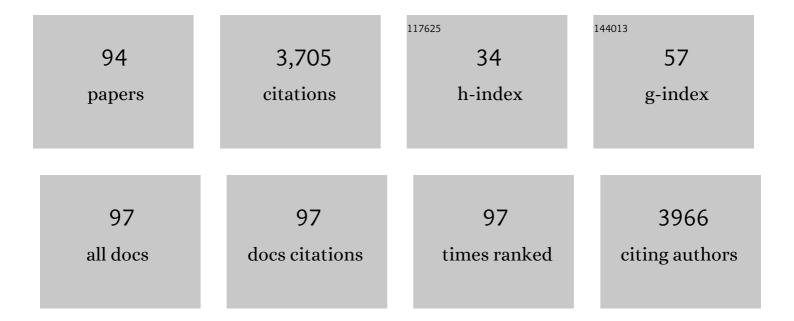
List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	SPP1 genotype is a determinant of disease severity in Duchenne muscular dystrophy. Neurology, 2011, 76, 219-226.	1.1	194
2	Reliability of the North Star Ambulatory Assessment in a multicentric setting. Neuromuscular Disorders, 2009, 19, 458-461.	0.6	171
3	North Star Ambulatory Assessment, 6-minute walk test and timed items in ambulant boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 2010, 20, 712-716.	0.6	171
4	Functional changes in Duchenne muscular dystrophy. Neurology, 2011, 77, 250-256.	1.1	151
5	Diagnostic and Prognostic Biomarkers in Amyotrophic Lateral Sclerosis. JAMA Neurology, 2017, 74, 525.	9.0	139
6	Prednisone/prednisolone and deflazacort regimens in the CINRG Duchenne Natural History Study. Neurology, 2015, 85, 1048-1055.	1.1	138
7	<i>DMD</i> genotypes and loss of ambulation in the CINRG Duchenne Natural History Study. Neurology, 2016, 87, 401-409.	1.1	119
8	Genetic modifiers of ambulation in the cooperative international Neuromuscular research group Duchenne natural history study. Annals of Neurology, 2015, 77, 684-696.	5.3	111
9	Discovery of serum protein biomarkers in the mdx mouse model and cross-species comparison to Duchenne muscular dystrophy patients. Human Molecular Genetics, 2014, 23, 6458-6469.	2.9	106
10	Nusinersen safety and effects on motor function in adult spinal muscular atrophy type 2 and 3. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 1166-1174.	1.9	99
11	24 Month Longitudinal Data in Ambulant Boys with Duchenne Muscular Dystrophy. PLoS ONE, 2013, 8, e52512.	2.5	99
12	Reliability of the Performance of Upper Limb assessment in Duchenne muscular dystrophy. Neuromuscular Disorders, 2014, 24, 201-206.	0.6	83
13	Importance of <i>SPP1</i> genotype as a covariate in clinical trials in Duchenne muscular dystrophy. Neurology, 2012, 79, 159-162.	1.1	81
14	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
15	Association Study of Exon Variants in the NF-κB and TGFβ Pathways Identifies CD40 as a Modifier of Duchenne Muscular Dystrophy. American Journal of Human Genetics, 2016, 99, 1163-1171.	6.2	71
16	Interpreting Genetic Variants in Titin in Patients With Muscle Disorders. JAMA Neurology, 2018, 75, 557.	9.0	69
17	Redefining phenotypes associated with mitochondrial DNA single deletion. Journal of Neurology, 2015, 262, 1301-1309.	3.6	68
18	6 Minute Walk Test in Duchenne MD Patients with Different Mutations: 12 Month Changes. PLoS ONE, 2014, 9, e83400.	2.5	65

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19	TNF-α-Induced microRNAs Control Dystrophin Expression in Becker Muscular Dystrophy. Cell Reports, 2015, 12, 1678-1690.	6.4	62
20	Non-neural phenotype of spinal and bulbar muscular atrophy: results from a large cohort of Italian patients. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 810-816.	1.9	59
21	Evidence for ACTN3 as a genetic modifier of Duchenne muscular dystrophy. Nature Communications, 2017, 8, 14143.	12.8	58
22	Genetic Modifiers of Duchenne Muscular Dystrophy and Dilated Cardiomyopathy. PLoS ONE, 2015, 10, e0141240.	2.5	58
23	The "Usual Suspects― Genes for Inflammation, Fibrosis, Regeneration, and Muscle Strength Modify Duchenne Muscular Dystrophy. Journal of Clinical Medicine, 2019, 8, 649.	2.4	55
24	A Mutation in the <i>CASQ1</i> Gene Causes a Vacuolar Myopathy with Accumulation of Sarcoplasmic Reticulum Protein Aggregates. Human Mutation, 2014, 35, 1163-1170.	2.5	53
25	Clinical and molecular characterization of limbâ€girdle muscular dystrophy due to <i>LAMA2</i> mutations. Muscle and Nerve, 2011, 44, 703-709.	2.2	52
26	Clinical and genetic spectrum in limb-girdle muscular dystrophy type 2E. Neurology, 2015, 84, 1772-1781.	1.1	50
27	Pilot trial of clenbuterol in spinal and bulbar muscular atrophy. Neurology, 2013, 80, 2095-2098.	1.1	47
28	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.	2.5	47
29	Upper limb function in Duchenne muscular dystrophy: 24 month longitudinal data. PLoS ONE, 2018, 13, e0199223.	2.5	45
30	New genotype-phenotype correlations in a large European cohort of patients with sarcoglycanopathy. Brain, 2020, 143, 2696-2708.	7.6	45
31	European muscle MRI study in limb girdle muscular dystrophy type R1/2A (LGMDR1/LGMD2A). Journal of Neurology, 2020, 267, 45-56.	3.6	43
32	Effect of Different Corticosteroid Dosing Regimens on Clinical Outcomes in Boys With Duchenne Muscular Dystrophy. JAMA - Journal of the American Medical Association, 2022, 327, 1456.	7.4	43
33	Benefits of glucocorticoids in non-ambulant boys/men with Duchenne muscular dystrophy: A multicentric longitudinal study using the Performance of Upper Limb test. Neuromuscular Disorders, 2015, 25, 749-753.	0.6	41
34	"l have got something positive out of this situation― psychological benefits of caregiving in relatives of young people with muscular dystrophy. Journal of Neurology, 2014, 261, 188-195.	3.6	37
35	Functional changes in Becker muscular dystrophy: implications for clinical trials in dystrophinopathies. Scientific Reports, 2016, 6, 32439.	3.3	36
36	TCTEX1D1 is a genetic modifier of disease progression in Duchenne muscular dystrophy. European Journal of Human Genetics, 2020, 28, 815-825.	2.8	36

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37	Genetic modifiers of respiratory function in Duchenne muscular dystrophy. Annals of Clinical and Translational Neurology, 2020, 7, 786-798.	3.7	36
38	Burden, professional support, and social network in families of children and young adults with muscular dystrophies. Muscle and Nerve, 2015, 52, 13-21.	2.2	35
39	Muscle MRI and functional outcome measures in Becker muscular dystrophy. Scientific Reports, 2017, 7, 16060.	3.3	35
40	Timed Rise from Floor as a Predictor of Disease Progression in Duchenne Muscular Dystrophy: An Observational Study. PLoS ONE, 2016, 11, e0151445.	2.5	32
41	Revisiting mitochondrial ocular myopathies: a study from the Italian Network. Journal of Neurology, 2017, 264, 1777-1784.	3.6	32
42	Cardiomyopathy in patients with POMT1-related congenital and limb-girdle muscular dystrophy. European Journal of Human Genetics, 2012, 20, 1234-1239.	2.8	31
43	Cognitive profile and MRI findings in limb-girdle muscular dystrophy 2I. Journal of Neurology, 2011, 258, 1312-1320.	3.6	25
44	Targeted gene panel screening is an effective tool to identify undiagnosed late onset Pompe disease. Neuromuscular Disorders, 2018, 28, 586-591.	0.6	24
45	The 6 Minute Walk Test and Performance of Upper Limb in Ambulant Duchenne Muscular Dystrophy Boys. PLOS Currents, 2014, 6, .	1.4	24
46	Psychological and practical difficulties among parents and healthy siblings of children with Duchenne vs. Becker muscular dystrophy: an Italian comparative study. Acta Myologica, 2014, 33, 136-43.	1.5	24
47	SPP1 genotype and glucocorticoid treatment modify osteopontin expression in Duchenne muscular dystrophy cells. Human Molecular Genetics, 2017, 26, 3342-3351.	2.9	23
48	<i>TGFBR2</i> but not <i>SPP1</i> genotype modulates osteopontin expression in Duchenne muscular dystrophy muscle. Journal of Pathology, 2012, 228, 251-259.	4.5	22
49	Whole genome sequencing reveals a 7 base-pair deletion in DMD exon 42 in a dog with muscular dystrophy. Mammalian Genome, 2017, 28, 106-113.	2.2	22
50	Genetic diagnosis as a tool for personalized treatment of Duchenne muscular dystrophy. Acta Myologica, 2016, 35, 122-127.	1.5	22
51	VBP15, a Novel Anti-Inflammatory, is Effective at Reducing the Severity of Murine Experimental Autoimmune Encephalomyelitis. Cellular and Molecular Neurobiology, 2015, 35, 377-387.	3.3	21
52	Assessment of disease progression in dysferlinopathy. Neurology, 2019, 92, .	1.1	20
53	Heterozygous frameshift variants in HNRNPA2B1 cause early-onset oculopharyngeal muscular dystrophy. Nature Communications, 2022, 13, 2306.	12.8	20
54	OPNâ€a induces muscle inflammation by increasing recruitment and activation of proâ€inflammatory macrophages. Experimental Physiology, 2016, 101, 1285-1300.	2.0	19

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55	Teenage exercise is associated with earlier symptom onset in dysferlinopathy: a retrospective cohort study. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 1224-1226.	1.9	19
56	Lipomatosis Incidence and Characteristics in an Italian Cohort of Mitochondrial Patients. Frontiers in Neurology, 2019, 10, 160.	2.4	19
57	Cardiorespiratory management of Duchenne muscular dystrophy: emerging therapies, neuromuscular genetics, and new clinical challenges. Lancet Respiratory Medicine,the, 2022, 10, 403-420.	10.7	19
58	Parkinson-like features in ALS with predominant upper motor neuron involvement. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2012, 13, 137-143.	2.1	18
59	Assessment of patients with idiopathic inflammatory myopathies and isolated creatin-kinase elevation. Autoimmunity Highlights, 2014, 5, 87-94.	3.9	18
60	Assessing Dysferlinopathy Patients Over Three Years With a New Motor Scale. Annals of Neurology, 2021, 89, 967-978.	5.3	17
61	Safety and efficacy of edaravone compared to historical controls in patients with amyotrophic lateral sclerosis from North-Eastern Italy. Journal of the Neurological Sciences, 2019, 404, 47-51.	0.6	16
62	DGUOK recessive mutations in patients with CPEO, mitochondrial myopathy, parkinsonism and mtDNA deletions. Brain, 2018, 141, e3-e3.	7.6	15
63	The clinical spectrum of CASQ1-related myopathy. Neurology, 2018, 91, e1629-e1641.	1.1	14
64	Teaching an Old Molecule New Tricks: Drug Repositioning for Duchenne Muscular Dystrophy. International Journal of Molecular Sciences, 2019, 20, 6053.	4.1	14
65	Osteopontin is linked with AKT, FoxO1, and myostatin in skeletal muscle cells. Muscle and Nerve, 2017, 56, 1119-1127.	2.2	12
66	Evaluation of mexiletine effect on conduction delay and bradyarrhythmic complications in patients with myotonic dystrophy type 1 over long-term follow-up. Heart Rhythm, 2020, 17, 1944-1950.	0.7	12
67	Changes in Muscle Metabolism are Associated with Phenotypic Variability in Golden Retriever Muscular Dystrophy. Yale Journal of Biology and Medicine, 2017, 90, 351-360.	0.2	12
68	Evaluation of peripherin in biofluids of patients with motor neuron diseases. Annals of Clinical and Translational Neurology, 2021, 8, 1750-1754.	3.7	11
69	Clinical and genetic spectrum of a large cohort of patients with δ-sarcoglycan muscular dystrophy. Brain, 2022, 145, 596-606.	7.6	11
70	Validation of the Italian version of the SBMA Functional Rating Scale as outcome measure. Neurological Sciences, 2016, 37, 1815-1821.	1.9	9
71	No effect of <i><scp>AR</scp></i> polyG polymorphism on spinal and bulbar muscular atrophy phenotype. European Journal of Neurology, 2016, 23, 1134-1136.	3.3	8
72	Influence of β2 adrenergic receptor genotype on risk of nocturnal ventilation in patients with Duchenne muscular dystrophy. Respiratory Research, 2019, 20, 221.	3.6	8

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73	Movement Disorders in Children with a Mitochondrial Disease: A Cross-Sectional Survey from the Nationwide Italian Collaborative Network of Mitochondrial Diseases. Journal of Clinical Medicine, 2021, 10, 2063.	2.4	8
74	The relevance of migraine in the clinical spectrum of mitochondrial disorders. Scientific Reports, 2022, 12, 4222.	3.3	7
75	Therapeutic opportunities and clinical outcome measures in Duchenne muscular dystrophy. Neurological Sciences, 2022, 43, 625-633.	1.9	7
76	Wholeâ€Body Muscle Magnetic Resonance Imaging in Glycogenâ€Storage Disease Type III. Muscle and Nerve, 2019, 60, 72-79.	2.2	6
77	North Star Ambulatory Assessment changes in ambulant Duchenne boys amenable to skip exons 44, 45, 51, and 53: A 3 year follow up. PLoS ONE, 2021, 16, e0253882.	2.5	6
78	RNA-seq in DMD urinary stem cells recognized muscle-related transcription signatures and addressed the identification of atypical mutations by whole-genome sequencing. Human Genetics and Genomics Advances, 2022, 3, 100054.	1.7	6
79	Integrated care of muscular dystrophies in Italy. Part 2. Psychological treatments, social and welfare support, and financial costs. Acta Myologica, 2017, 36, 41-45.	1.5	6
80	Quality of life assessment in adult spinal muscular atrophy patients treated with nusinersen. Journal of Neurology, 2022, 269, 3264-3275.	3.6	6
81	Becker muscular dystrophy due to an intronic splicing mutation inducing a dual dystrophin transcript. Neuromuscular Disorders, 2016, 26, 662-665.	0.6	5
82	Ablation of collagen VI leads to the release of platelets with altered function. Blood Advances, 2021, 5, 5150-5163.	5.2	5
83	Integrated care of muscular dystrophies in Italy. Part 1. Pharmacological treatment and rehabilitative interventions. Acta Myologica, 2017, 36, 19-24.	1.5	4
84	Assessing the Relationship of Patient Reported Outcome Measures With Functional Status in Dysferlinopathy: A Rasch Analysis Approach. Frontiers in Neurology, 2022, 13, 828525.	2.4	4
85	The Role of Motor System in Mental Rotation: New Insights from Myotonic Dystrophy Type 1. Journal of the International Neuropsychological Society, 2020, 26, 492-502.	1.8	3
86	Whole-body muscle MRI in McArdle disease. Neuromuscular Disorders, 2022, 32, 5-14.	0.6	3
87	Influence of β2 adrenergic receptor genotype on longitudinal measures of forced vital capacity in patients with Duchenne muscular dystrophy. Neuromuscular Disorders, 2022, 32, 150-158.	0.6	3
88	Muscle MR Imaging in Tubular Aggregate Myopathy. PLoS ONE, 2014, 9, e94427.	2.5	2
89	Genome-Wide Association Studies in Muscle Physiology and Disease. , 2019, , 9-30.		2
90	Limb girdle muscular dystrophy due to gene mutations: new mutations expand the clinical spectrum of a still challenging diagnosis. Acta Myologica, 2020, 39, 67-82.	1.5	2

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91	Genetic modifiers of upper limb function in Duchenne muscular dystrophy. Journal of Neurology, 2022, 269, 4884-4894.	3.6	2
92	Longitudinal motor function in proximal versus distal <scp><i>DMD</i></scp> pathogenic variants. Muscle and Nerve, 2021, 64, 467-473.	2.2	1
93	Predicting age at loss of ambulation in Duchenne muscular dystrophy with deep phenotypic measures. , 2014, , .		0
94	The nonsense mutation stop+4 model correlates with motor changes in Duchenne muscular dystrophy. Neuromuscular Disorders, 2021, 31, 479-488.	0.6	0