Luca Bello

List of Publications by Year
in descending order

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94 papers	3,705 citations	117625 34 h-index	144013 57 g-index
97 all docs	97 docs citations	97 times ranked	3966 citing authors

#	Article	IF	Citations
1	Whole-body muscle MRI in McArdle disease. Neuromuscular Disorders, 2022, 32, 5-14.	0.6	3
2	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
3	Clinical and genetic spectrum of a large cohort of patients with Î´-sarcoglycan muscular dystrophy. Brain, 2022, 145, 596-606.	7.6	11
4	Influence of Î²2 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
5	Quality of life assessment in adult spinal muscular atrophy patients treated with nusinersen. Journal of Neurology, 2022, 269, 3264-3275.	3.6	6
6	The relevance of migraine in the clinical spectrum of mitochondrial disorders. Scientific Reports, 2022, 12, 4222.	3.3	7
7	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
8	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
9	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
10	Heterozygous frameshift variants in HNRNPA2B1 cause early-onset oculopharyngeal muscular dystrophy. Nature Communications, 2022, 13, 2306.	12.8	20
11	Genetic modifiers of upper limb function in Duchenne muscular dystrophy. Journal of Neurology, 2022, 269, 4884-4894.	3.6	2
12	Therapeutic opportunities and clinical outcome measures in Duchenne muscular dystrophy. Neurological Sciences, 2022, 43, 625-633.	1.9	7
13	Assessing Dysferlinopathy Patients Over Three Years With a New Motor Scale. Annals of Neurology, 2021, 89, 967-978.	5.3	17
14	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
15	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
16	The nonsense mutation stop+4 model correlates with motor changes in Duchenne muscular dystrophy. Neuromuscular Disorders, 2021, 31, 479-488.	0.6	0
17	Longitudinal motor function in proximal versus distal <scp>DMD</scp> pathogenic variants. Muscle and Nerve, 2021, 64, 467-473.	2.2	1
18	Evaluation of peripherin in biofluids of patients with motor neuron diseases. Annals of Clinical and Translational Neurology, 2021, 8, 1750-1754.	3.7	11

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19	Ablation of collagen VI leads to the release of platelets with altered function. Blood Advances, 2021, 5, 5150-5163.	5.2	5
20	European muscle MRI study in limb girdle muscular dystrophy type R1/2A (LGMDR1/LGMD2A). Journal of Neurology, 2020, 267, 45-56.	3.6	43
21	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
22	TCTEX1D1 is a genetic modifier of disease progression in Duchenne muscular dystrophy. European Journal of Human Genetics, 2020, 28, 815-825.	2.8	36
23	New genotype-phenotype correlations in a large European cohort of patients with sarcoglycanopathy. Brain, 2020, 143, 2696-2708.	7.6	45
24	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
25	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
26	Genetic modifiers of respiratory function in Duchenne muscular dystrophy. Annals of Clinical and Translational Neurology, 2020, 7, 786-798.	3.7	36
27	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
28	Influence of Î²2 adrenergic receptor genotype on risk of nocturnal ventilation in patients with Duchenne muscular dystrophy. Respiratory Research, 2019, 20, 221.	3.6	8
29	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
30	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
31	The â€œUsual Suspectsâ€• Genes for Inflammation, Fibrosis, Regeneration, and Muscle Strength Modify Duchenne Muscular Dystrophy. Journal of Clinical Medicine, 2019, 8, 649.	2.4	55
32	Lipomatosis Incidence and Characteristics in an Italian Cohort of Mitochondrial Patients. Frontiers in Neurology, 2019, 10, 160.	2.4	19
33	Wholeâ€Body Muscle Magnetic Resonance Imaging in Glycogenâ€Storage Disease Type III. Muscle and Nerve, 2019, 60, 72-79.	2.2	6
34	Teaching an Old Molecule New Tricks: Drug Repositioning for Duchenne Muscular Dystrophy. International Journal of Molecular Sciences, 2019, 20, 6053.	4.1	14
35	Assessment of disease progression in dysferlinopathy. Neurology, 2019, 92, .	1.1	20
36	Genome-Wide Association Studies in Muscle Physiology and Disease. , 2019, , 9-30.		2

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37	Targeted gene panel screening is an effective tool to identify undiagnosed late onset Pompe disease. Neuromuscular Disorders, 2018, 28, 586-591.	0.6	24
38	Interpreting Genetic Variants in Titin in Patients With Muscle Disorders. JAMA Neurology, 2018, 75, 557.	9.0	69
39	DGUOK recessive mutations in patients with CPEO, mitochondrial myopathy, parkinsonism and mtDNA deletions. Brain, 2018, 141, e3-e3.	7.6	15
40	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
41	The clinical spectrum of CASQ1-related myopathy. Neurology, 2018, 91, e1629-e1641.	1.1	14
42	Upper limb function in Duchenne muscular dystrophy: 24 month longitudinal data. PLoS ONE, 2018, 13, e0199223.	2.5	45
43	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
44	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
45	Evidence for ACTN3 as a genetic modifier of Duchenne muscular dystrophy. Nature Communications, 2017, 8, 14143.	12.8	58
46	Diagnostic and Prognostic Biomarkers in Amyotrophic Lateral Sclerosis. JAMA Neurology, 2017, 74, 525.	9.0	139
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	Osteopontin is linked with AKT, FoxO1, and myostatin in skeletal muscle cells. Muscle and Nerve, 2017, 56, 1119-1127.	2.2	12
49	Muscle MRI and functional outcome measures in Becker muscular dystrophy. Scientific Reports, 2017, 7, 16060.	3.3	35
50	Revisiting mitochondrial ocular myopathies: a study from the Italian Network. Journal of Neurology, 2017, 264, 1777-1784.	3.6	32
51	Integrated care of muscular dystrophies in Italy. Part 1. Pharmacological treatment and rehabilitative interventions. Acta Myologica, 2017, 36, 19-24.	1.5	4
52	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
53	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
54	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

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55	No effect of <scp>AR</scp> polyG polymorphism on spinal and bulbar muscular atrophy phenotype. European Journal of Neurology, 2016, 23, 1134-1136.	3.3	8
56	OPNâ€a induces muscle inflammation by increasing recruitment and activation of proâ€inflammatory macrophages. Experimental Physiology, 2016, 101, 1285-1300.	2.0	19
57	Validation of the Italian version of the SBMA Functional Rating Scale as outcome measure. Neurological Sciences, 2016, 37, 1815-1821.	1.9	9
58	Becker muscular dystrophy due to an intronic splicing mutation inducing a dual dystrophin transcript. Neuromuscular Disorders, 2016, 26, 662-665.	0.6	5
59	DMD genotypes and loss of ambulation in the CINRG Duchenne Natural History Study. Neurology, 2016, 87, 401-409.	1.1	119
60	Association Study of Exon Variants in the NF-ÎºB and TGFÎ² Pathways Identifies CD40 as a Modifier of Duchenne Muscular Dystrophy. American Journal of Human Genetics, 2016, 99, 1163-1171.	6.2	71
61	Functional changes in Becker muscular dystrophy: implications for clinical trials in dystrophinopathies. Scientific Reports, 2016, 6, 32439.	3.3	36
62	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
63	Genetic diagnosis as a tool for personalized treatment of Duchenne muscular dystrophy. Acta Myologica, 2016, 35, 122-127.	1.5	22
64	TNF-Î±-Induced microRNAs Control Dystrophin Expression in Becker Muscular Dystrophy. Cell Reports, 2015, 12, 1678-1690.	6.4	62
65	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
66	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
67	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
68	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
69	Clinical and genetic spectrum in limb-girdle muscular dystrophy type 2E. Neurology, 2015, 84, 1772-1781.	1.1	50
70	Redefining phenotypes associated with mitochondrial DNA single deletion. Journal of Neurology, 2015, 262, 1301-1309.	3.6	68
71	Prednisone/prednisolone and deflazacort regimens in the CINRG Duchenne Natural History Study. Neurology, 2015, 85, 1048-1055.	1.1	138
72	Genetic Modifiers of Duchenne Muscular Dystrophy and Dilated Cardiomyopathy. PLoS ONE, 2015, 10, e0141240.	2.5	58

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73	Muscle MR Imaging in Tubular Aggregate Myopathy. PLoS ONE, 2014, 9, e94427.	2.5	2
74	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
75	Assessment of patients with idiopathic inflammatory myopathies and isolated creatin-kinase elevation. Autoimmunity Highlights, 2014, 5, 87-94.	3.9	18
76	6 Minute Walk Test in Duchenne MD Patients with Different Mutations: 12 Month Changes. PLoS ONE, 2014, 9, e83400.	2.5	65
77	Reliability of the Performance of Upper Limb assessment in Duchenne muscular dystrophy. Neuromuscular Disorders, 2014, 24, 201-206.	0.6	83
78	â€œI have got something positive out of this situationâ€• psychological benefits of caregiving in relatives of young people with muscular dystrophy. Journal of Neurology, 2014, 261, 188-195.	3.6	37
79	A Mutation in theCASQ1Gene Causes a Vacuolar Myopathy with Accumulation of Sarcoplasmic Reticulum Protein Aggregates. Human Mutation, 2014, 35, 1163-1170.	2.5	53
80	Predicting age at loss of ambulation in Duchenne muscular dystrophy with deep phenotypic measures. , 2014, , .		0
81	The 6 Minute Walk Test and Performance of Upper Limb in Ambulant Duchenne Muscular Dystrophy Boys. PLOS Currents, 2014, 6, .	1.4	24
82	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
83	Pilot trial of clenbuterol in spinal and bulbar muscular atrophy. Neurology, 2013, 80, 2095-2098.	1.1	47
84	24 Month Longitudinal Data in Ambulant Boys with Duchenne Muscular Dystrophy. PLoS ONE, 2013, 8, e52512.	2.5	99
85	Cardiomyopathy in patients with POMT1-related congenital and limb-girdle muscular dystrophy. European Journal of Human Genetics, 2012, 20, 1234-1239.	2.8	31
86	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
87	Importance of SPP1 genotype as a covariate in clinical trials in Duchenne muscular dystrophy. Neurology, 2012, 79, 159-162.	1.1	81
88	TGFBR2 but not SPP1 genotype modulates osteopontin expression in Duchenne muscular dystrophy muscle. Journal of Pathology, 2012, 228, 251-259.	4.5	22
89	Cognitive profile and MRI findings in limb-girdle muscular dystrophy 2I. Journal of Neurology, 2011, 258, 1312-1320.	3.6	25
90	Clinical and molecular characterization of limbâ€girdle muscular dystrophy due to LAMA2 mutations. Muscle and Nerve, 2011, 44, 703-709.	2.2	52

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91	Functional changes in Duchenne muscular dystrophy. Neurology, 2011, 77, 250-256.	1.1	151
92	SPP1 genotype is a determinant of disease severity in Duchenne muscular dystrophy. Neurology, 2011, 76, 219-226.	1.1	194
93	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
94	Reliability of the North Star Ambulatory Assessment in a multicentric setting. Neuromuscular Disorders, 2009, 19, 458-461.	0.6	171