Elena Pegoraro

List of Publications by Year in descending order

76 papers	2,918 citations	136885 32 h-index	182361 51 g-index
79 all docs	79 docs citations	79 times ranked	3278 citing authors

#	Article	IF	Citations
1	Quality of life assessment in adult spinal muscular atrophy patients treated with nusinersen. Journal of Neurology, 2022, 269, 3264-3275.	1.8	6
2	Cardiac and pulmonary findings in dysferlinopathy: A 3â€year, longitudinal study. Muscle and Nerve, 2022, 65, 531-540.	1.0	9
3	The relevance of migraine in the clinical spectrum of mitochondrial disorders. Scientific Reports, 2022, 12, 4222.	1.6	7
4	Assessing the Relationship of Patient Reported Outcome Measures With Functional Status in Dysferlinopathy: A Rasch Analysis Approach. Frontiers in Neurology, 2022, 13, 828525.	1.1	4
5	Cardiorespiratory management of Duchenne muscular dystrophy: emerging therapies, neuromuscular genetics, and new clinical challenges. Lancet Respiratory Medicine,the, 2022, 10, 403-420.	5.2	19
6	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.	3.8	43
7	Expanding the clinical-pathological and genetic spectrum of RYR1-related congenital myopathies with cores and minicores: an Italian population study. Acta Neuropathologica Communications, 2022, 10, 54.	2.4	3
8	Cognitive profiles and clinical factors in type III spinal muscular atrophy: a preliminary study. Neuromuscular Disorders, 2022, 32, 672-677.	0.3	5
9	Genetic modifiers of upper limb function in Duchenne muscular dystrophy. Journal of Neurology, 2022, 269, 4884-4894.	1.8	2
10	Nutrition in adult patients with selected lysosomal storage diseases. Nutrition, Metabolism and Cardiovascular Diseases, 2021, 31, 733-744.	1.1	7
11	Assessing Dysferlinopathy Patients Over Three Years With a New Motor Scale. Annals of Neurology, 2021, 89, 967-978.	2.8	17
12	Efficacy and Safety of Bimagrumab in Sporadic Inclusion Body Myositis. Neurology, 2021, 96, e1595-e1607.	1.5	25
13	Miyoshi myopathy and limb girdle muscular dystrophy R2 are the same disease. Neuromuscular Disorders, 2021, 31, 265-280.	0.3	18
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.	1.0	8
15	A 14-Year Italian Experience in DM2 Genetic Testing: Frequency and Distribution of Normal and Premutated CNBP Alleles. Frontiers in Genetics, 2021, 12, 668094.	1.1	3
16	De novo revertant fiber formation and therapy testing in a 3D culture model of Duchenne muscular dystrophy skeletal muscle. Acta Biomaterialia, 2021, 132, 227-244.	4.1	26
17	European muscle MRI study in limb girdle muscular dystrophy type R1/2A (LGMDR1/LGMD2A). Journal of Neurology, 2020, 267, 45-56.	1.8	43
18	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.	1.2	3

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19	Outcome measures and treatment effectiveness in late onset myasthenia gravis. Neurological Research and Practice, 2020, 2, 45.	1.0	8
20	New genotype-phenotype correlations in a large European cohort of patients with sarcoglycanopathy. Brain, 2020, 143, 2696-2708.	3.7	45
21	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.	0.3	12
22	Recognition of emotions conveyed by facial expression and body postures in myotonic dystrophy (DM). Cortex, 2020, 127, 58-66.	1.1	19
23	Genetic modifiers of respiratory function in Duchenne muscular dystrophy. Annals of Clinical and Translational Neurology, 2020, 7, 786-798.	1.7	36
24	Intensive Teenage Activity Is Associated With Greater Muscle Hyperintensity on T1W Magnetic Resonance Imaging in Adults With Dysferlinopathy. Frontiers in Neurology, 2020, 11, 613446.	1.1	3
25	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
26	238th ENMC International Workshop: Updating management recommendations of cardiac dystrophinopathyHoofddorp, The Netherlands, 30 November - 2 December 2018. Neuromuscular Disorders, 2019, 29, 634-643.	0.3	6
27	A 3D culture model of innervated human skeletal muscle enables studies of the adult neuromuscular junction. ELife, 2019, 8, .	2.8	169
28	The â€œUsual Suspectsâ€• Genes for Inflammation, Fibrosis, Regeneration, and Muscle Strength Modify Duchenne Muscular Dystrophy. Journal of Clinical Medicine, 2019, 8, 649.	1.0	55
29	Cored in the act: the use of models to understand core myopathies. DMM Disease Models and Mechanisms, 2019, 12, .	1.2	17
30	DGUOK recessive mutations in patients with CPEO, mitochondrial myopathy, parkinsonism and mtDNA deletions. Brain, 2018, 141, e3-e3.	3.7	15
31	Inositol trisphosphate receptor-mediated Ca2+ signalling stimulates mitochondrial function and gene expression in core myopathy patients. Human Molecular Genetics, 2018, 27, 2367-2382.	1.4	14
32	Teenage exercise is associated with earlier symptom onset in dysferlinopathy: a retrospective cohort study. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 1224-1226.	0.9	19
33	MRI in sarcoglycanopathies: a large international cohort study. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 72-77.	0.9	55
34	The clinical spectrum of CASQ1-related myopathy. Neurology, 2018, 91, e1629-e1641.	1.5	14
35	Upper limb function in Duchenne muscular dystrophy: 24 month longitudinal data. PLoS ONE, 2018, 13, e0199223.	1.1	45
36	Muscle MRI in patients with dysferlinopathy: pattern recognition and implications for clinical trials. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 1071-1081.	0.9	81

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37	Evidence for ACTN3 as a genetic modifier of Duchenne muscular dystrophy. Nature Communications, 2017, 8, 14143.	5.8	58
38	The role of transmission electron microscopy in vacuole-associated myopathies. Ultrastructural Pathology, 2017, 41, 88-90.	0.4	1
39	SPP1 genotype and glucocorticoid treatment modify osteopontin expression in Duchenne muscular dystrophy cells. Human Molecular Genetics, 2017, 26, 3342-3351.	1.4	23
40	Muscle MRI and functional outcome measures in Becker muscular dystrophy. Scientific Reports, 2017, 7, 16060.	1.6	35
41	Association Study of Exon Variants in the NF-Î°B and TGFÎ² Pathways Identifies CD40 as a Modifier of Duchenne Muscular Dystrophy. American Journal of Human Genetics, 2016, 99, 1163-1171.	2.6	71
42	Functional changes in Becker muscular dystrophy: implications for clinical trials in dystrophinopathies. Scientific Reports, 2016, 6, 32439.	1.6	36
43	The genetic basis of undiagnosed muscular dystrophies and myopathies. Neurology, 2016, 87, 71-76.	1.5	92
44	Reliable and versatile immortal muscle cell models from healthy and myotonic dystrophy type 1 primary human myoblasts. Experimental Cell Research, 2016, 342, 39-51.	1.2	32
45	Clinical and molecular study in a long-surviving patient with MLASA syndrome due to novel PUS1 mutations. Neurogenetics, 2016, 17, 65-70.	0.7	29
46	Genetic diagnosis as a tool for personalized treatment of Duchenne muscular dystrophy. Acta Myologica, 2016, 35, 122-127.	1.5	22
47	Genetic modifiers of ambulation in the cooperative international Neuromuscular research group Duchenne natural history study. Annals of Neurology, 2015, 77, 684-696.	2.8	111
48	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.	0.3	41
49	Clinical and genetic spectrum in limb-girdle muscular dystrophy type 2E. Neurology, 2015, 84, 1772-1781.	1.5	50
50	A Diagnostic Dilemma in a Family With Cystinuria Type B Resolved by Muscle Magnetic Resonance. Pediatric Neurology, 2015, 52, 548-551.	1.0	4
51	Centronuclear myopathies: genotypeâ€“phenotype correlation and frequency of defined genetic forms in an Italian cohort. Journal of Neurology, 2015, 262, 1728-1740.	1.8	51
52	Genetic Modifiers of Duchenne Muscular Dystrophy and Dilated Cardiomyopathy. PLoS ONE, 2015, 10, e0141240.	1.1	58
53	6 Minute Walk Test in Duchenne MD Patients with Different Mutations: 12 Month Changes. PLoS ONE, 2014, 9, e83400.	1.1	65
54	Reliability of the Performance of Upper Limb assessment in Duchenne muscular dystrophy. Neuromuscular Disorders, 2014, 24, 201-206.	0.3	83

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55	A Mutation in theCASQ1Gene Causes a Vacuolar Myopathy with Accumulation of Sarcoplasmic Reticulum Protein Aggregates. Human Mutation, 2014, 35, 1163-1170.	1.1	53
56	The 6 Minute Walk Test and Performance of Upper Limb in Ambulant Duchenne Muscular Dystrophy Boys. PLOS Currents, 2014, 6, .	1.4	24
57	Androgen-dependent impairment of myogenesis in spinal and bulbar muscular atrophy. Acta Neuropathologica, 2013, 126, 109-121.	3.9	41
58	Alterations in Osteopontin Modify Muscle Size in Females in Both Humans and Mice. Medicine and Science in Sports and Exercise, 2013, 45, 1060-1068.	0.2	35
59	Importance of SPP1 genotype as a covariate in clinical trials in Duchenne muscular dystrophy. Neurology, 2012, 79, 159-162.	1.5	81
60	TGFBR2 but not SPP1 genotype modulates osteopontin expression in Duchenne muscular dystrophy muscle. Journal of Pathology, 2012, 228, 251-259.	2.1	22
61	Clinical and molecular characterization of limbâ€girdle muscular dystrophy due to LAMA2 mutations. Muscle and Nerve, 2011, 44, 703-709.	1.0	52
62	MYH7 gene mutation in myosin storage myopathy and scapulo-peroneal myopathy. Neuromuscular Disorders, 2007, 17, 321-329.	0.3	46
63	Biochemical and ultrastructural evidence of endoplasmic reticulum stress in LGMD2I. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2007, 451, 1047-1055.	1.4	23
64	A novel out-of-frame mutation in the neurofilament light chain gene (NEFL) does not result in Charcot-Marie-Tooth disease type 2E. Neurogenetics, 2005, 6, 49-50.	0.7	5
65	Clinical and Molecular Characterization of Patients With Limb-Girdle Muscular Dystrophy Type 2I. Archives of Neurology, 2005, 62, 1894.	4.9	81
66	Decorin and biglycan expression is differentially altered in several muscular dystrophies. Brain, 2005, 128, 2546-2555.	3.7	87
67	Co-segregation of LMNA and PMP22 gene mutations in the same family. Neuromuscular Disorders, 2005, 15, 858-862.	0.3	12
68	Clinical and molecular study in congenital muscular dystrophy with partial laminin ?2 (LAMA2) deficiency. Human Mutation, 2003, 21, 103-111.	1.1	49
69	X-inactivation pattern in multiple tissues from two leber's hereditary optic neuropathy (LHON) patients. American Journal of Medical Genetics Part A, 2003, 119A, 37-40.	2.4	21
70	Integrin Î±7Î²1 in Muscular Dystrophy/Myopathy of Unknown Etiology. American Journal of Pathology, 2002, 160, 2135-2143.	1.9	59
71	Familial Skewed X Inactivation: A Molecular Trait Associated with High Spontaneous-Abortion Rate Maps to Xq28. American Journal of Human Genetics, 1997, 61, 160-170.	2.6	132
72	Congenital muscular dystrophy with primary laminin ?2 (merosin) deficiency presenting as inflammatory myopathy. Annals of Neurology, 1996, 40, 782-791.	2.8	119

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73	X-inactivation patterns in female Leber's hereditary optic neuropathy patients do not support a strong X-linked determinant. , 1996, 61, 356-362.		36
74	Genetic counseling of isolated carriers of Duchenne muscular dystrophy. , 1996, 63, 573-580.		28
75	Xâ€inactivation patterns in female Leber's hereditary optic neuropathy patients do not support a strong Xâ€linked determinant. American Journal of Medical Genetics Part A, 1996, 61, 356-362.	2.4	1
76	Myotonic dystrophy: evidence for a possible dominant-negative RNA mutation. Human Molecular Genetics, 1995, 4, 599-606.	1.4	179