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	Myotonic dystrophy: evidence for a possible dominant-negative RNA mutation. Human Molecular Genetics, 1995, 4, 599-606.	1.4	179
2	A 3D culture model of innervated human skeletal muscle enables studies of the adult neuromuscular junction. ELife, 2019, 8, .	2.8	169
3	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
4	Congenital muscular dystrophy with primary laminin ?2 (merosin) deficiency presenting as inflammatory myopathy. Annals of Neurology, 1996, 40, 782-791.	2.8	119
5	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
6	The genetic basis of undiagnosed muscular dystrophies and myopathies. Neurology, 2016, 87, 71-76.	1.5	92
7	Decorin and biglycan expression is differentially altered in several muscular dystrophies. Brain, 2005, 128, 2546-2555.	3.7	87
8	Reliability of the Performance of Upper Limb assessment in Duchenne muscular dystrophy. Neuromuscular Disorders, 2014, 24, 201-206.	0.3	83
9	Clinical and Molecular Characterization of Patients With Limb-Girdle Muscular Dystrophy Type 2I. Archives of Neurology, 2005, 62, 1894.	4.9	81
10	Importance of SPP1 genotype as a covariate in clinical trials in Duchenne muscular dystrophy. Neurology, 2012, 79, 159-162.	1.5	81
11	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
12	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.	2.6	71
13	6 Minute Walk Test in Duchenne MD Patients with Different Mutations: 12 Month Changes. PLoS ONE, 2014, 9, e83400.	1.1	65
14	Integrin Î±7Î²1 in Muscular Dystrophy/Myopathy of Unknown Etiology. American Journal of Pathology, 2002, 160, 2135-2143.	1.9	59
15	Evidence for ACTN3 as a genetic modifier of Duchenne muscular dystrophy. Nature Communications, 2017, 8, 14143.	5.8	58
16	Genetic Modifiers of Duchenne Muscular Dystrophy and Dilated Cardiomyopathy. PLoS ONE, 2015, 10, e0141240.	1.1	58
17	MRI in sarcoglycanopathies: a large international cohort study. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 72-77.	0.9	55
18	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

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19	A Mutation in theCASQ1Gene Causes a Vacuolar Myopathy with Accumulation of Sarcoplasmic Reticulum Protein Aggregates. Human Mutation, 2014, 35, 1163-1170.	1.1	53
20	Clinical and molecular characterization of limbâ€girdle muscular dystrophy due to LAMA2 mutations. Muscle and Nerve, 2011, 44, 703-709.	1.0	52
21	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
22	Clinical and genetic spectrum in limb-girdle muscular dystrophy type 2E. Neurology, 2015, 84, 1772-1781.	1.5	50
23	Clinical and molecular study in congenital muscular dystrophy with partial laminin ?2 (LAMA2) deficiency. Human Mutation, 2003, 21, 103-111.	1.1	49
24	MYH7 gene mutation in myosin storage myopathy and scapulo-peroneal myopathy. Neuromuscular Disorders, 2007, 17, 321-329.	0.3	46
25	Upper limb function in Duchenne muscular dystrophy: 24 month longitudinal data. PLoS ONE, 2018, 13, e0199223.	1.1	45
26	New genotype-phenotype correlations in a large European cohort of patients with sarcoglycanopathy. Brain, 2020, 143, 2696-2708.	3.7	45
27	European muscle MRI study in limb girdle muscular dystrophy type R1/2A (LGMDR1/LGMD2A). Journal of Neurology, 2020, 267, 45-56.	1.8	43
28	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
29	Androgen-dependent impairment of myogenesis in spinal and bulbar muscular atrophy. Acta Neuropathologica, 2013, 126, 109-121.	3.9	41
30	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
31	X-inactivation patterns in female Leber's hereditary optic neuropathy patients do not support a strong X-linked determinant. , 1996, 61, 356-362.		36
32	Functional changes in Becker muscular dystrophy: implications for clinical trials in dystrophinopathies. Scientific Reports, 2016, 6, 32439.	1.6	36
33	Genetic modifiers of respiratory function in Duchenne muscular dystrophy. Annals of Clinical and Translational Neurology, 2020, 7, 786-798.	1.7	36
34	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
35	Muscle MRI and functional outcome measures in Becker muscular dystrophy. Scientific Reports, 2017, 7, 16060.	1.6	35
36	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

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37	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
38	Genetic counseling of isolated carriers of Duchenne muscular dystrophy. , 1996, 63, 573-580.		28
39	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
40	Efficacy and Safety of Bimagrumab in Sporadic Inclusion Body Myositis. Neurology, 2021, 96, e1595-e1607.	1.5	25
41	The 6 Minute Walk Test and Performance of Upper Limb in Ambulant Duchenne Muscular Dystrophy Boys. PLOS Currents, 2014, 6, .	1.4	24
42	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
43	SPP1 genotype and glucocorticoid treatment modify osteopontin expression in Duchenne muscular dystrophy cells. Human Molecular Genetics, 2017, 26, 3342-3351.	1.4	23
44	TGFBR2 but not SPP1 genotype modulates osteopontin expression in Duchenne muscular dystrophy muscle. Journal of Pathology, 2012, 228, 251-259.	2.1	22
45	Genetic diagnosis as a tool for personalized treatment of Duchenne muscular dystrophy. Acta Myologica, 2016, 35, 122-127.	1.5	22
46	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
47	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
48	Recognition of emotions conveyed by facial expression and body postures in myotonic dystrophy (DM). Cortex, 2020, 127, 58-66.	1.1	19
49	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
50	Miyoshi myopathy and limb girdle muscular dystrophy R2 are the same disease. Neuromuscular Disorders, 2021, 31, 265-280.	0.3	18
51	Cored in the act: the use of models to understand core myopathies. DMM Disease Models and Mechanisms, 2019, 12, .	1.2	17
52	Assessing Dysferlinopathy Patients Over Three Years With a New Motor Scale. Annals of Neurology, 2021, 89, 967-978.	2.8	17
53	DGUOK recessive mutations in patients with CPEO, mitochondrial myopathy, parkinsonism and mtDNA deletions. Brain, 2018, 141, e3-e3.	3.7	15
54	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

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55	The clinical spectrum of CASQ1-related myopathy. Neurology, 2018, 91, e1629-e1641.	1.5	14
56	Co-segregation of LMNA and PMP22 gene mutations in the same family. Neuromuscular Disorders, 2005, 15, 858-862.	0.3	12
57	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
58	Cardiac and pulmonary findings in dysferlinopathy: A 3â€year, longitudinal study. Muscle and Nerve, 2022, 65, 531-540.	1.0	9
59	Outcome measures and treatment effectiveness in late onset myasthenia gravis. Neurological Research and Practice, 2020, 2, 45.	1.0	8
60	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
61	Nutrition in adult patients with selected lysosomal storage diseases. Nutrition, Metabolism and Cardiovascular Diseases, 2021, 31, 733-744.	1.1	7
62	The relevance of migraine in the clinical spectrum of mitochondrial disorders. Scientific Reports, 2022, 12, 4222.	1.6	7
63	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
64	Quality of life assessment in adult spinal muscular atrophy patients treated with nusinersen. Journal of Neurology, 2022, 269, 3264-3275.	1.8	6
65	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
66	Cognitive profiles and clinical factors in type III spinal muscular atrophy: a preliminary study. Neuromuscular Disorders, 2022, 32, 672-677.	0.3	5
67	A Diagnostic Dilemma in a Family With Cystinuria Type B Resolved by Muscle Magnetic Resonance. Pediatric Neurology, 2015, 52, 548-551.	1.0	4
68	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
69	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
70	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
71	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
72	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

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73	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
74	Genetic modifiers of upper limb function in Duchenne muscular dystrophy. Journal of Neurology, 2022, 269, 4884-4894.	1.8	2
75	The role of transmission electron microscopy in vacuole-associated myopathies. Ultrastructural Pathology, 2017, 41, 88-90.	0.4	1
76	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