Francesca Magri

List of Publications by Year
in descending order

85 papers	3,810 citations	117625 34 h-index	128289 60 g-index
86 all docs	86 docs citations	86 times ranked	5259 citing authors

#	Article	IF	Citations
1	Ataluren treatment of patients with nonsense mutation dystrophinopathy. Muscle and Nerve, 2014, 50, 477-487.	2.2	357
2	Genetic Correction of Human Induced Pluripotent Stem Cells from Patients with Spinal Muscular Atrophy. Science Translational Medicine, 2012, 4, 165ra162.	12.4	180
3	Reliability of the North Star Ambulatory Assessment in a multicentric setting. Neuromuscular Disorders, 2009, 19, 458-461.	0.6	171
4	Clinical, molecular, and protein correlations in a large sample of genetically diagnosed Italian limb girdle muscular dystrophy patients. Human Mutation, 2008, 29, 258-266.	2.5	162
5	Histological effects of givinostat in boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 2016, 26, 643-649.	0.6	144
6	Direct reprogramming of human astrocytes into neural stem cells and neurons. Experimental Cell Research, 2012, 318, 1528-1541.	2.6	143
7	Genotype and phenotype characterization in a large dystrophinopathic cohort with extended follow-up. Journal of Neurology, 2011, 258, 1610-1623.	3.6	134
8	Mutations in DNA2 Link Progressive Myopathy to Mitochondrial DNA Instability. American Journal of Human Genetics, 2013, 92, 293-300.	6.2	115
9	SOD1 misplacing and mitochondrial dysfunction in amyotrophic lateral sclerosis pathogenesis. Frontiers in Cellular Neuroscience, 2015, 9, 336.	3.7	111
10	24 Month Longitudinal Data in Ambulant Boys with Duchenne Muscular Dystrophy. PLoS ONE, 2013, 8, e52512.	2.5	99
11	Long Term Natural History Data in Ambulant Boys with Duchenne Muscular Dystrophy: 36-Month Changes. PLoS ONE, 2014, 9, e108205.	2.5	98
12	The genetic basis of undiagnosed muscular dystrophies and myopathies. Neurology, 2016, 87, 71-76.	1.1	92
13	The italian limb girdle muscular dystrophy registry: Relative frequency, clinical features, and differential diagnosis. Muscle and Nerve, 2017, 55, 55-68.	2.2	86
14	Importance of SPP1 genotype as a covariate in clinical trials in Duchenne muscular dystrophy. Neurology, 2012, 79, 159-162.	1.1	81
15	Next-generation sequencing reveals DGUOK mutations in adult patients with mitochondrial DNA multiple deletions. Brain, 2012, 135, 3404-3415.	7.6	81
16	MotorPlex provides accurate variant detection across large muscle genes both in single myopathic patients and in pools of DNA samples. Acta Neuropathologica Communications, 2014, 2, 100.	5.2	76
17	Prevalence of congenital muscular dystrophy in Italy. Neurology, 2015, 84, 904-911.	1.1	75
18	Therapeutic Development in Amyotrophic Lateral Sclerosis. Clinical Therapeutics, 2015, 37, 668-680.	2.5	71

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19	Interpreting Genetic Variants in Titin in Patients With Muscle Disorders. JAMA Neurology, 2018, 75, 557.	9.0	69
20	Mutation finding in patients with dysferlin deficiency and role of the dysferlin interacting proteins annexin A1 and A2 in muscular dystrophies. Human Mutation, 2005, 26, 283-283.	2.5	65
21	6 Minute Walk Test in Duchenne MD Patients with Different Mutations: 12 Month Changes. PLoS ONE, 2014, 9, e83400.	2.5	65
22	Genetic Modifiers of Duchenne Muscular Dystrophy and Dilated Cardiomyopathy. PLoS ONE, 2015, 10, e0141240.	2.5	58
23	Categorizing natural history trajectories of ambulatory function measured by the 6-minute walk distance in patients with Duchenne muscular dystrophy. Neuromuscular Disorders, 2016, 26, 576-583.	0.6	57
24	Frequency and characterisation of anoctamin 5 mutations in a cohort of Italian limb-girdle muscular dystrophy patients. Neuromuscular Disorders, 2012, 22, 934-943.	0.6	53
25	Ongoing therapeutic trials and outcome measures for Duchenne muscular dystrophy. Cellular and Molecular Life Sciences, 2013, 70, 4585-4602.	5.4	53
26	Centronuclear myopathies: genotypeâ€“phenotype correlation and frequency of defined genetic forms in an Italian cohort. Journal of Neurology, 2015, 262, 1728-1740.	3.6	51
27	Molecular etiopathogenesis of limb girdle muscular and congenital muscular dystrophies: Boundaries and contiguities. Clinica Chimica Acta, 2005, 361, 54-79.	1.1	48
28	Antisense Oligonucleotide Therapy for the Treatment of C9ORF72 ALS/FTD Diseases. Molecular Neurobiology, 2014, 50, 721-732.	4.0	48
29	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
30	Nusinersen treatment and cerebrospinal fluid neurofilaments: An explorative study on Spinal Muscular Atrophy type 3 patients. Journal of Cellular and Molecular Medicine, 2020, 24, 3034-3039.	3.6	47
31	Neurocognitive Profiles in Duchenne Muscular Dystrophy and Gene Mutation Site. Pediatric Neurology, 2011, 45, 292-299.	2.1	46
32	mi<scp>RNA</scp> in spinal muscular atrophy pathogenesis and therapy. Journal of Cellular and Molecular Medicine, 2018, 22, 755-767.	3.6	46
33	Nitric oxide donor and non steroidal anti inflammatory drugs as a therapy for muscular dystrophies: Evidence from a safety study with pilot efficacy measures in adult dystrophic patients. Pharmacological Research, 2012, 65, 472-479.	7.1	40
34	Histologic muscular history in steroid-treated and untreated patients with Duchenne dystrophy. Neurology, 2015, 85, 1886-1893.	1.1	39
35	Multiparametric quantitative MRI assessment of thigh muscles in limbâ€girdle muscular dystrophy 2A and 2B. Muscle and Nerve, 2018, 58, 550-558.	2.2	37
36	Genetic modifiers of respiratory function in Duchenne muscular dystrophy. Annals of Clinical and Translational Neurology, 2020, 7, 786-798.	3.7	36

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37	Respiratory pattern in an adult population of dystrophic patients. Journal of the Neurological Sciences, 2011, 306, 54-61.	0.6	35
38	Clinical and molecular characterization of a cohort of patients with novel nucleotide alterations of the Dystrophin gene detected by direct sequencing. BMC Medical Genetics, 2011, 12, 37.	2.1	32
39	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
40	Human induced pluripotent stem cell models for the study and treatment of Duchenne and Becker muscular dystrophies. Therapeutic Advances in Neurological Disorders, 2019, 12, 175628641983347.	3.5	32
41	The wide spectrum of clinical phenotypes of spinal muscular atrophy with respiratory distress type 1: A systematic review. Journal of the Neurological Sciences, 2014, 346, 35-42.	0.6	30
42	Pluripotent stem cell-based models of spinal muscular atrophy. Molecular and Cellular Neurosciences, 2015, 64, 44-50.	2.2	28
43	Incontinence in Late-Onset Pompe Disease: An Underdiagnosed Treatable Condition. European Neurology, 2012, 68, 75-78.	1.4	27
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	Estimating the impact of COVID-19 pandemic on services provided by Italian Neuromuscular Centers: an Italian Association of Myology survey of the acute phase. Acta Myologica, 2020, 39, 57-66.	1.5	24
46	New molecular findings in congenital myopathies due to selenoprotein N gene mutations. Journal of the Neurological Sciences, 2011, 300, 107-113.	0.6	23
47	Extended phenotype description and new molecular findings in late onset glycogen storage disease type II: a northern Italy population study and review of the literature. Journal of Neurology, 2014, 261, 83-97.	3.6	23
48	Myotonia congenita: Novel mutations in CLCN1 gene and functional characterizations in Italian patients. Journal of the Neurological Sciences, 2012, 318, 65-71.	0.6	22
49	Optic atrophy plus phenotype due to mutations in the OPA1 gene: Two more Italian families. Journal of the Neurological Sciences, 2012, 315, 146-149.	0.6	21
50	Stormorken Syndrome Caused by a p.R304W STIM1 Mutation: The First Italian Patient and a Review of the Literature. Frontiers in Neurology, 2018, 9, 859.	2.4	20
51	Research advances in gene therapy approaches for the treatment of amyotrophic lateral sclerosis. Cellular and Molecular Life Sciences, 2012, 69, 1641-1650.	5.4	19
52	New Mutations in NEB Gene Discovered by Targeted Next-Generation Sequencing in Nemaline Myopathy Italian Patients. Journal of Molecular Neuroscience, 2016, 59, 351-359.	2.3	17
53	Longitudinal follow-up and muscle MRI pattern of two siblings with polyglucosan body myopathy due to glycogenin-1 mutation. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 797-800.	1.9	17
54	Can Intestinal Pseudo-Obstruction Drive Recurrent Stroke-Like Episodes in Late-Onset MELAS Syndrome? A Case Report and Review of the Literature. Frontiers in Neurology, 2019, 10, 38.	2.4	17

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55	Adult Polyglucosan Body Disease: Clinical and histological heterogeneity of a large Italian family. Neuromuscular Disorders, 2015, 25, 423-428.	0.6	14
56	Postural effects on lung and chest wall volumes in late onset type II glycogenosis patients. Respiratory Physiology and Neurobiology, 2013, 186, 308-314.	1.6	13
57	Noncoding RNAs in Duchenne and Becker muscular dystrophies: role in pathogenesis and future prognostic and therapeutic perspectives. Cellular and Molecular Life Sciences, 2020, 77, 4299-4313.	5.4	13
58	Tyr78Phe Transthyretin Mutation with Predominant Motor Neuropathy as the Initial Presentation. Case Reports in Neurology, 2011, 3, 62-68.	0.7	12
59	A new case of limb girdle muscular dystrophy 2G in a Greek patient, founder effect and review of the literature. Neuromuscular Disorders, 2018, 28, 532-537.	0.6	11
60	Revised Genetic Classification of Limb Girdle Muscular Dystrophies. Current Molecular Medicine, 2014, 14, 934-943.	1.3	11
61	Muscle histological changes in a large cohort of patients affected with Becker muscular dystrophy. Acta Neuropathologica Communications, 2022, 10, 48.	5.2	11
62	ISPD mutations account for a small proportion of Italian Limb Girdle Muscular Dystrophy cases. BMC Neurology, 2015, 15, 172.	1.8	10
63	Diagnostic and prognostic value of CSF neurofilaments in a cohort of patients with motor neuron disease: A crossâ€sectional study. Journal of Cellular and Molecular Medicine, 2021, 25, 3765-3771.	3.6	10
64	In vitro analysis of splice site mutations in the CLCN1 gene using the minigene assay. Molecular Biology Reports, 2014, 41, 2865-2874.	2.3	8
65	Copy Number Variants Account for a Tiny Fraction of Undiagnosed Myopathic Patients. Genes, 2018, 9, 524.	2.4	7
66	Impact of <scp>COVIDâ€19</scp> on the quality of life of patients with neuromuscular disorders in the <scp>L</scp>ombardy area, <scp>I</scp>taly. Muscle and Nerve, 2021, 64, 474-482.	2.2	7
67	Central Nervous System Involvement in Common Variable Immunodeficiency: A Case of Acute Unilateral Optic Neuritis in a 26-Year-Old Italian Patient. Frontiers in Neurology, 2018, 9, 1031.	2.4	6
68	Bilateral Cavernous Carotid Aneurysms: Atypical Presentation of a Rare Cause of Mass Effect. A Case Report and a Review of the Literature. Frontiers in Neurology, 2018, 9, 619.	2.4	6
69	Missense mutations in small muscle protein X-linked (SMPX) cause distal myopathy with protein inclusions. Acta Neuropathologica, 2021, 142, 375-393.	7.7	6
70	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
71	Sodium Channel Myotonia Due to Novel Mutations in Domain I of Nav1.4. Frontiers in Neurology, 2020, 11, 255.	2.4	5
72	Ophthalmoplegia Due to Miller Fisher Syndrome in a Patient With Myasthenia Gravis. Frontiers in Neurology, 2019, 10, 823.	2.4	4

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73	Subclinical Leberâ€™s hereditary optic neuropathy with pediatric acute spinal cord onset: more than meets the eye. BMC Neurology, 2018, 18, 220.	1.8	3
74	Early Findings in Neonatal Cases of RYR1â€“Related Congenital Myopathies. Frontiers in Neurology, 2021, 12, 664618.	2.4	3
75	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.	5.2	3
76	G.P.251. Neuromuscular Disorders, 2014, 24, 892.	0.6	2
77	Anti-sulfatide reactivity in patients with celiac disease. Scandinavian Journal of Gastroenterology, 2017, 52, 409-413.	1.5	2
78	CACNA1S mutation associated with a case of juvenile-onset congenital myopathy. Journal of the Neurological Sciences, 2021, 431, 120047.	0.6	2
79	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
80	Genetic modifiers of upper limb function in Duchenne muscular dystrophy. Journal of Neurology, 2022, 269, 4884-4894.	3.6	2
81	G.P.7.05 Becker muscular dystrophy with a stop codon mutation in the 5â€² of the dystrophin gene. Neuromuscular Disorders, 2008, 18, 777-778.	0.6	1
82	P.2.7 6min walk test 12month changes in DMD: Correlation with genotype. Neuromuscular Disorders, 2013, 23, 750-751.	0.6	1
83	Spontaneous Hydromyelic Cavity in Two Unrelated Patients with Late-Onset Pompe Disease: Is This a Fortuitous Association?. European Neurology, 2013, 70, 102-105.	1.4	1
84	Expanding the clinical spectrum of the mitochondrial mutation A13084T in the ND5 gene. Neurology: Genetics, 2020, 6, e511.	1.9	1
85	The nonsense mutation stop+4 model correlates with motor changes in Duchenne muscular dystrophy. Neuromuscular Disorders, 2021, 31, 479-488.	0.6	0