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	Muscle histological changes in a large cohort of patients affected with Becker muscular dystrophy. Acta Neuropathologica Communications, 2022, 10, 48.	5.2	11
2	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
3	Genetic modifiers of upper limb function in Duchenne muscular dystrophy. Journal of Neurology, 2022, 269, 4884-4894.	3.6	2
4	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
5	Missense mutations in small muscle protein X-linked (SMPX) cause distal myopathy with protein inclusions. Acta Neuropathologica, 2021, 142, 375-393.	7.7	6
6	Early Findings in Neonatal Cases of RYR1â€"Related Congenital Myopathies. Frontiers in Neurology, 2021, 12, 664618.	2.4	3
7	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
8	The nonsense mutation stop+4 model correlates with motor changes in Duchenne muscular dystrophy. Neuromuscular Disorders, 2021, 31, 479-488.	0.6	0
9	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
10	CACNA1S mutation associated with a case of juvenile-onset congenital myopathy. Journal of the Neurological Sciences, 2021, 431, 120047.	0.6	2
11	Expanding the clinical spectrum of the mitochondrial mutation A13084T in the ND5 gene. Neurology: Genetics, 2020, 6, e511.	1.9	1
12	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
13	Genetic modifiers of respiratory function in Duchenne muscular dystrophy. Annals of Clinical and Translational Neurology, 2020, 7, 786-798.	3.7	36
14	Sodium Channel Myotonia Due to Novel Mutations in Domain I of Nav1.4. Frontiers in Neurology, 2020, 11, 255.	2.4	5
15	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
16	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
17	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
18	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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19	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
20	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
21	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
22	mi<scp>RNA</scp> in spinal muscular atrophy pathogenesis and therapy. Journal of Cellular and Molecular Medicine, 2018, 22, 755-767.	3.6	46
23	Targeted gene panel screening is an effective tool to identify undiagnosed late onset Pompe disease. Neuromuscular Disorders, 2018, 28, 586-591.	0.6	24
24	Interpreting Genetic Variants in Titin in Patients With Muscle Disorders. JAMA Neurology, 2018, 75, 557.	9.0	69
25	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
26	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
27	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
28	Copy Number Variants Account for a Tiny Fraction of Undiagnosed Myopathic Patients. Genes, 2018, 9, 524.	2.4	7
29	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
30	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
31	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
32	The italian limb girdle muscular dystrophy registry: Relative frequency, clinical features, and differential diagnosis. Muscle and Nerve, 2017, 55, 55-68.	2.2	86
33	Anti-sulfatide reactivity in patients with celiac disease. Scandinavian Journal of Gastroenterology, 2017, 52, 409-413.	1.5	2
34	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
35	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
36	Histological effects of givinostat in boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 2016, 26, 643-649.	0.6	144

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37	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
38	The genetic basis of undiagnosed muscular dystrophies and myopathies. Neurology, 2016, 87, 71-76.	1.1	92
39	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
40	ISPD mutations account for a small proportion of Italian Limb Girdle Muscular Dystrophy cases. BMC Neurology, 2015, 15, 172.	1.8	10
41	SOD1 misplacing and mitochondrial dysfunction in amyotrophic lateral sclerosis pathogenesis. Frontiers in Cellular Neuroscience, 2015, 9, 336.	3.7	111
42	Prevalence of congenital muscular dystrophy in Italy. Neurology, 2015, 84, 904-911.	1.1	75
43	Therapeutic Development in Amyotrophic Lateral Sclerosis. Clinical Therapeutics, 2015, 37, 668-680.	2.5	71
44	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
45	Adult Polyglucosan Body Disease: Clinical and histological heterogeneity of a large Italian family. Neuromuscular Disorders, 2015, 25, 423-428.	0.6	14
46	Histologic muscular history in steroid-treated and untreated patients with Duchenne dystrophy. Neurology, 2015, 85, 1886-1893.	1.1	39
47	Pluripotent stem cell-based models of spinal muscular atrophy. Molecular and Cellular Neurosciences, 2015, 64, 44-50.	2.2	28
48	Genetic Modifiers of Duchenne Muscular Dystrophy and Dilated Cardiomyopathy. PLoS ONE, 2015, 10, e0141240.	2.5	58
49	Long Term Natural History Data in Ambulant Boys with Duchenne Muscular Dystrophy: 36-Month Changes. PLoS ONE, 2014, 9, e108205.	2.5	98
50	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
51	Ataluren treatment of patients with nonsense mutation dystrophinopathy. Muscle and Nerve, 2014, 50, 477-487.	2.2	357
52	6 Minute Walk Test in Duchenne MD Patients with Different Mutations: 12 Month Changes. PLoS ONE, 2014, 9, e83400.	2.5	65
53	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
54	Antisense Oligonucleotide Therapy for the Treatment of C9ORF72 ALS/FTD Diseases. Molecular Neurobiology, 2014, 50, 721-732.	4.0	48

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55	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
56	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
57	G.P.251. Neuromuscular Disorders, 2014, 24, 892.	0.6	2
58	Revised Genetic Classification of Limb Girdle Muscular Dystrophies. Current Molecular Medicine, 2014, 14, 934-943.	1.3	11
59	Ongoing therapeutic trials and outcome measures for Duchenne muscular dystrophy. Cellular and Molecular Life Sciences, 2013, 70, 4585-4602.	5.4	53
60	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
61	Mutations in DNA2 Link Progressive Myopathy to Mitochondrial DNA Instability. American Journal of Human Genetics, 2013, 92, 293-300.	6.2	115
62	P.2.7 6min walk test 12month changes in DMD: Correlation with genotype. Neuromuscular Disorders, 2013, 23, 750-751.	0.6	1
63	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
64	24 Month Longitudinal Data in Ambulant Boys with Duchenne Muscular Dystrophy. PLoS ONE, 2013, 8, e52512.	2.5	99
65	Incontinence in Late-Onset Pompe Disease: An Underdiagnosed Treatable Condition. European Neurology, 2012, 68, 75-78.	1.4	27
66	Importance of SPP1 genotype as a covariate in clinical trials in Duchenne muscular dystrophy. Neurology, 2012, 79, 159-162.	1.1	81
67	Next-generation sequencing reveals DGUOK mutations in adult patients with mitochondrial DNA multiple deletions. Brain, 2012, 135, 3404-3415.	7.6	81
68	Genetic Correction of Human Induced Pluripotent Stem Cells from Patients with Spinal Muscular Atrophy. Science Translational Medicine, 2012, 4, 165ra162.	12.4	180
69	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
70	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
71	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
72	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

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73	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
74	Direct reprogramming of human astrocytes into neural stem cells and neurons. Experimental Cell Research, 2012, 318, 1528-1541.	2.6	143
75	New molecular findings in congenital myopathies due to selenoprotein N gene mutations. Journal of the Neurological Sciences, 2011, 300, 107-113.	0.6	23
76	Respiratory pattern in an adult population of dystrophic patients. Journal of the Neurological Sciences, 2011, 306, 54-61.	0.6	35
77	Neurocognitive Profiles in Duchenne Muscular Dystrophy and Gene Mutation Site. Pediatric Neurology, 2011, 45, 292-299.	2.1	46
78	Genotype and phenotype characterization in a large dystrophinopathic cohort with extended follow-up. Journal of Neurology, 2011, 258, 1610-1623.	3.6	134
79	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
80	Tyr78Phe Transthyretin Mutation with Predominant Motor Neuropathy as the Initial Presentation. Case Reports in Neurology, 2011, 3, 62-68.	0.7	12
81	Reliability of the North Star Ambulatory Assessment in a multicentric setting. Neuromuscular Disorders, 2009, 19, 458-461.	0.6	171
82	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
83	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
84	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
85	Molecular etiopathogenesis of limb girdle muscular and congenital muscular dystrophies: Boundaries and contiguities. Clinica Chimica Acta, 2005, 361, 54-79.	1.1	48