Giulia Ricci

List of Publications by Year
in descending order

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81	2,326	186265	233421
papers	citations	28	45
		h-index	g-index

82	82	82	3575
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Astrocyteâ€“neuron interactions in neurological disorders. Journal of Biological Physics, 2009, 35, 317-336.	1.5	144
2	Mitochondria and Neurodegeneration. Bioscience Reports, 2007, 27, 87-104.	2.4	125
3	Large-Scale Population Analysis Challenges the Current Criteria for the Molecular Diagnosis of Fascioscapulohumeral Muscular Dystrophy. American Journal of Human Genetics, 2012, 90, 628-635.	6.2	104
4	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
5	The genetic basis of undiagnosed muscular dystrophies and myopathies. Neurology, 2016, 87, 71-76.	1.1	92
6	The italian limb girdle muscular dystrophy registry: Relative frequency, clinical features, and differential diagnosis. Muscle and Nerve, 2017, 55, 55-68.	2.2	86
7	Large scale genotypeâ€“phenotype analyses indicate that novel prognostic tools are required for families with facioscapulohumeral muscular dystrophy. Brain, 2013, 136, 3408-3417.	7.6	85
8	Mitochondrial DNA transcription and translation: clinical syndromes. Essays in Biochemistry, 2018, 62, 321-340.	4.7	72
9	Next-Generation Sequencing Identifies Transportin 3 as the Causative Gene for LGMD1F. PLoS ONE, 2013, 8, e63536.	2.5	69
10	Genetic characterization in symptomatic female DMD carriers: lack of relationship between X-inactivation, transcriptional DMD allele balancing and phenotype. BMC Medical Genetics, 2012, 13, 73.	2.1	63
11	Disease awareness in myotonic dystrophy type 1: an observational cross-sectional study. Orphanet Journal of Rare Diseases, 2016, 11, 34.	2.7	63
12	Folate, Homocysteine, Vitamin B12, and Polymorphisms of Genes Participating in One-Carbon Metabolism in Late-Onset Alzheimer's Disease Patients and Healthy Controls. Antioxidants and Redox Signaling, 2012, 17, 195-204.	5.4	60
13	Clinical expression of facioscapulohumeral muscular dystrophy in carriers of 1â€“3 D4Z4 reduced alleles: experience of the FSHD Italian National Registry. BMJ Open, 2016, 6, e007798.	1.9	60
14	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
15	LMNA -associated myopathies. Neurology, 2014, 83, 1634-1644.	1.1	57
16	New motor outcome function measures in evaluation of Lateâ€Onset Pompe disease before and after enzyme replacement therapy. Muscle and Nerve, 2012, 45, 831-834.	2.2	56
17	A novel clinical tool to classify facioscapulohumeral muscular dystrophy phenotypes. Journal of Neurology, 2016, 263, 1204-1214.	3.6	55
18	Facioscapulohumeral muscular dystrophy: new insights from compound heterozygotes and implication for prenatal genetic counselling. Journal of Medical Genetics, 2012, 49, 171-178.	3.2	53

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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.	2.5	53
20	Identification and characterization of three novel mutations in theCASQ1gene in four patients with tubular aggregate myopathy. Human Mutation, 2017, 38, 1761-1773.	2.5	51
21	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
22	Functional Diagnostics in Mitochondrial Diseases. Bioscience Reports, 2007, 27, 53-67.	2.4	43
23	Lack of association between mtDNA haplogroups and Alzheimerâ€™s disease in Tuscany. Neurological Sciences, 2007, 28, 142-147.	1.9	41
24	Oxidative Stress Treatment for Clinical Trials in Neurodegenerative Diseases. Journal of Alzheimer's Disease, 2011, 24, 111-126.	2.6	39
25	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
26	Cardiac and Neuromuscular Features of Patients WithLMNA-Related Cardiomyopathy. Annals of Internal Medicine, 2019, 171, 458.	3.9	33
27	Effects of aerobic training on exercise-related oxidative stress in mitochondrial myopathies. Neuromuscular Disorders, 2012, 22, S172-S177.	0.6	31
28	Rippling muscle disease and facioscapulohumeral dystrophy-like phenotype in a patient carrying a heterozygous CAV3 T78M mutation and a D4Z4 partial deletion: Further evidence for â€œdouble troubleâ€• overlapping syndromes. Neuromuscular Disorders, 2012, 22, 534-540.	0.6	28
29	Autosomal dominant psychiatric disorders and mitochondrial DNA multiple deletions: Report of a family. Journal of Affective Disorders, 2008, 106, 173-177.	4.1	27
30	Mutations in glycyl-tRNA synthetase impair mitochondrial metabolism in neurons. Human Molecular Genetics, 2018, 27, 2187-2204.	2.9	26
31	Elevated TGF Î²2 serum levels in Emery-Dreifuss Muscular Dystrophy: Implications for myocyte and tenocyte differentiation and fibrogenic processes. Nucleus, 2018, 9, 337-349.	2.2	25
32	Phenotypic Variability Among Patients With D4Z4 Reduced Allele Facioscapulohumeral Muscular Dystrophy. JAMA Network Open, 2020, 3, e204040.	5.9	25
33	ClCâ€1 mutations in myotonia congenita patients: insights into molecular gating mechanisms and genotypeâ€“phenotype correlation. Journal of Physiology, 2015, 593, 4181-4199.	2.9	24
34	Multidisciplinary study of a new ClCâ€1 mutation causing myotonia congenita: a paradigm to understand and treat ion channelopathies. FASEB Journal, 2016, 30, 3285-3295.	0.5	24
35	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
36	Muscle exercise in limb girdle muscular dystrophies: pitfall and advantages. Acta Myologica, 2015, 34, 3-8.	1.5	20

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37	Interpretation of the Epigenetic Signature of Facioscapulohumeral Muscular Dystrophy in Light of Genotype-Phenotype Studies. International Journal of Molecular Sciences, 2020, 21, 2635.	4.1	18
38	Large genotypeâ€“phenotype study in carriers of D4Z4 borderline alleles provides guidance for facioscapulohumeral muscular dystrophy diagnosis. Scientific Reports, 2020, 10, 21648.	3.3	16
39	Metabolic myopathies: functional evaluation by different exercise testing approaches. Musculoskeletal Surgery, 2011, 95, 59-67.	1.5	15
40	A mobile app for patients with Pompe disease and its possible clinical applications. Neuromuscular Disorders, 2018, 28, 471-475.	0.6	15
41	A 5-year clinical follow-up study from the Italian National Registry for FSHD. Journal of Neurology, 2021, 268, 356-366.	3.6	15
42	An â€œinflammatoryâ€•mitochondrial myopathy. A case report. Neuromuscular Disorders, 2013, 23, 907-910.	0.6	13
43	Next-generation sequencing application to investigate skeletal muscle channelopathies in a large cohort of Italian patients. Neuromuscular Disorders, 2021, 31, 336-347.	0.6	13
44	Prevalent cardiac phenotype resulting in heart transplantation in a novel LMNA gene duplication. Neuromuscular Disorders, 2010, 20, 512-516.	0.6	12
45	Phenotype may predict the clinical course of facioscapolohumeral muscular dystrophy. Muscle and Nerve, 2019, 59, 711-713.	2.2	12
46	Central Nervous System Involvement as Outcome Measure for Clinical Trials Efficacy in Myotonic Dystrophy Type 1. Frontiers in Neurology, 2020, 11, 624.	2.4	12
47	Pes cavus and hereditary neuropathies: when a relationship should be suspected. Journal of Orthopaedics and Traumatology, 2010, 11, 195-201.	2.3	11
48	A personal monitoring architecture to detect muscular fatigue in elderly. Neuromuscular Disorders, 2012, 22, S192-S197.	0.6	11
49	Cardiac magnetic resonance in patients with muscular dystrophies. European Journal of Preventive Cardiology, 2021, 28, 1526-1535.	1.8	11
50	Exercise-Related Oxidative Stress as Mechanism to Fight Physical Dysfunction in Neuromuscular Disorders. Frontiers in Physiology, 2020, 11, 451.	2.8	11
51	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
52	An Ontology-Driven Multisensorial Platform to Enable Unobtrusive Human Monitoring and Independent Living. , 2009, , .		8
53	Serum gamma-glutamyltransferase fractions in Myotonic Dystrophy type I: Differences with healthy subjects and patients with liver disease. Clinical Biochemistry, 2010, 43, 1246-1248.	1.9	8
54	Assessing the Role of Anti rh-GAA in Modulating Response to ERT in a Late-Onset Pompe Disease Cohort from the Italian GSDII Study Group. Advances in Therapy, 2019, 36, 1177-1189.	2.9	8

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55	Cytokine Profile in Striated Muscle Laminopathies: New Promising Biomarkers for Disease Prediction. Cells, 2020, 9, 1532.	4.1	8
56	Psychosocial impact of sport activity in neuromuscular disorders. Neurological Sciences, 2020, 41, 2561-2567.	1.9	8
57	A wearable pervasive platform for the intelligent monitoring of muscular fatigue. , 2010, , .		7
58	Nerve, muscle and heart acute toxicity following oxaliplatin and capecitabine treatment. Neuromuscular Disorders, 2012, 22, 767-770.	0.6	7
59	The empowerment of translational research: lessons from laminopathies. Orphanet Journal of Rare Diseases, 2012, 7, 37.	2.7	7
60	A novel family with axonal Charcotâ€Marieâ€Tooth disease caused by a mutation in the EGR2 gene. Journal of the Peripheral Nervous System, 2019, 24, 219-223.	3.1	7
61	Clinical and Molecular Spectrum of Myotonia and Periodic Paralyses Associated With Mutations in SCN4A in a Large Cohort of Italian Patients. Frontiers in Neurology, 2020, 11, 646.	2.4	7
62	Increased resistance towards fatigability in patients with facioscapulohumeral muscular dystrophy. European Journal of Applied Physiology, 2021, 121, 1617-1629.	2.5	7
63	Expanding the clinical and genetic spectrum of pathogenic variants in <scp>STIM1</scp>. Muscle and Nerve, 2021, 64, 567-575.	2.2	7
64	Therapeutic opportunities and clinical outcome measures in Duchenne muscular dystrophy. Neurological Sciences, 2022, 43, 625-633.	1.9	7
65	A multi-parametric protocol to study exercise intolerance in McArdle's disease. Acta Myologica, 2015, 34, 120-125.	1.5	6
66	Exercise therapy in muscle diseases: open issues and future perspectives. Acta Myologica, 2019, 38, 233-238.	1.5	6
67	Integrated care of muscular dystrophies in Italy. Part 1. Pharmacological treatment and rehabilitative interventions. Acta Myologica, 2017, 36, 19-24.	1.5	4
68	Fatigue as a common signature of inflammatory myopathies: clinical aspects and care.. Clinical and Experimental Rheumatology, 2022, 40, 425-432.	0.8	4
69	A pervasive activity management and rehabilitation support system for the elderly. , 2010, , .		3
70	A case of intravascular large B cell lymphoma with brain involvement mimicking progressive multifocal leukoencephalopathy. International Journal of Neuroscience, 2023, 133, 735-739.	1.6	2
71	Hard ways towards adulthood: the transition phase in young people with myotonic dystrophy. Acta Myologica, 2016, 35, 145-149.	1.5	2
72	Neuromuscular tetanic hyperexcitability syndrome associated to a heterozygous mutation with normal serum magnesium levels. Acta Myologica, 2020, 39, 36-39.	1.5	2

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73	A Single mtDNA Deletion in Association with a LMNA Gene New Frameshift Variant: A Case Report. Journal of Neuromuscular Diseases, 2022, 9, 457-462.	2.6	2
74	Inflammatory myopathy in a patient with postural and kinetik tremor. Neurological Sciences, 2011, 32, 1175-1178.	1.9	1
75	Posterior reversible encephalopathy syndrome in a complicated autoimmune background: differential diagnosis and etiological hypothesis. Acta Neurologica Belgica, 2013, 113, 191-193.	1.1	1
76	Anti-HMGCR antibodies and asymptomatic hyperCKemia. A case report. Acta Myologica, 2021, 40, 105-108.	1.5	1
77	Frailties and critical issues in neuromuscular diseases highlighted by SARS-CoV-2 pandemic: how many patients are still "invisible"?. Acta Myologica, 2022, 41, 24-29.	1.5	1
78	Unusual concomitant smallâ€•and largeâ€fiber neuropathy related to hypereosinophilic syndrome. Clinical and Experimental Neuroimmunology, 0, , .	1.0	0
79	Management of motor rehabilitation in individuals with muscular dystrophies. 1 Consensus Conference report from UILDM - Italian Muscular Dystrophy Association (Rome, January 25-26, 2019). Acta Myologica, 2021, 40, 72-87.	1.5	0
80	31â€ƒCardiac magnetic resonance findings in patients with Type 1 myotonic dystrophy. European Heart Journal Supplements, 2021, 23, .	0.1	0
81	Fatigue as a common signature of inflammatory myopathies: clinical aspects and care.. Clinical and Experimental Rheumatology, 2022, 40, 425-432.	0.8	0