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	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
2	Fatigue as a common signature of inflammatory myopathies: clinical aspects and care.. Clinical and Experimental Rheumatology, 2022, 40, 425-432.	0.8	4
3	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
4	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
5	Fatigue as a common signature of inflammatory myopathies: clinical aspects and care.. Clinical and Experimental Rheumatology, 2022, 40, 425-432.	0.8	0
6	Therapeutic opportunities and clinical outcome measures in Duchenne muscular dystrophy. Neurological Sciences, 2022, 43, 625-633.	1.9	7
7	Cardiac magnetic resonance in patients with muscular dystrophies. European Journal of Preventive Cardiology, 2021, 28, 1526-1535.	1.8	11
8	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
9	A 5-year clinical follow-up study from the Italian National Registry for FSHD. Journal of Neurology, 2021, 268, 356-366.	3.6	15
10	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
11	Increased resistance towards fatigability in patients with facioscapulohumeral muscular dystrophy. European Journal of Applied Physiology, 2021, 121, 1617-1629.	2.5	7
12	Expanding the clinical and genetic spectrum of pathogenic variants in <scp>STIM1</scp>. Muscle and Nerve, 2021, 64, 567-575.	2.2	7
13	Anti-HMGCR antibodies and asymptomatic hyperCKemia. A case report. Acta Myologica, 2021, 40, 105-108.	1.5	1
14	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
15	31â€ƒCardiac magnetic resonance findings in patients with Type 1 myotonic dystrophy. European Heart Journal Supplements, 2021, 23, .	0.1	0
16	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
17	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
18	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

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19	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
20	Phenotypic Variability Among Patients With D4Z4 Reduced Allele Facioscapulohumeral Muscular Dystrophy. JAMA Network Open, 2020, 3, e204040.	5.9	25
21	Exercise-Related Oxidative Stress as Mechanism to Fight Physical Dysfunction in Neuromuscular Disorders. Frontiers in Physiology, 2020, 11, 451.	2.8	11
22	Cytokine Profile in Striated Muscle Laminopathies: New Promising Biomarkers for Disease Prediction. Cells, 2020, 9, 1532.	4.1	8
23	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
24	Psychosocial impact of sport activity in neuromuscular disorders. Neurological Sciences, 2020, 41, 2561-2567.	1.9	8
25	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
26	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
27	Neuromuscular tetanic hyperexcitability syndrome associated to a heterozygous mutation with normal serum magnesium levels. Acta Myologica, 2020, 39, 36-39.	1.5	2
28	Phenotype may predict the clinical course of facioscapolohumeral muscular dystrophy. Muscle and Nerve, 2019, 59, 711-713.	2.2	12
29	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
30	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
31	Cardiac and Neuromuscular Features of Patients WithLMNA-Related Cardiomyopathy. Annals of Internal Medicine, 2019, 171, 458.	3.9	33
32	Exercise therapy in muscle diseases: open issues and future perspectives. Acta Myologica, 2019, 38, 233-238.	1.5	6
33	A mobile app for patients with Pompe disease and its possible clinical applications. Neuromuscular Disorders, 2018, 28, 471-475.	0.6	15
34	Mutations in glycyl-tRNA synthetase impair mitochondrial metabolism in neurons. Human Molecular Genetics, 2018, 27, 2187-2204.	2.9	26
35	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
36	Mitochondrial DNA transcription and translation: clinical syndromes. Essays in Biochemistry, 2018, 62, 321-340.	4.7	72

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37	The italian limb girdle muscular dystrophy registry: Relative frequency, clinical features, and differential diagnosis. Muscle and Nerve, 2017, 55, 55-68.	2.2	86
38	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
39	Integrated care of muscular dystrophies in Italy. Part 1. Pharmacological treatment and rehabilitative interventions. Acta Myologica, 2017, 36, 19-24.	1.5	4
40	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
41	A novel clinical tool to classify facioscapulohumeral muscular dystrophy phenotypes. Journal of Neurology, 2016, 263, 1204-1214.	3.6	55
42	The genetic basis of undiagnosed muscular dystrophies and myopathies. Neurology, 2016, 87, 71-76.	1.1	92
43	Multidisciplinary study of a new CICâ€1 mutation causing myotonia congenita: a paradigm to understand and treat ion channelopathies. FASEB Journal, 2016, 30, 3285-3295.	0.5	24
44	Disease awareness in myotonic dystrophy type 1: an observational cross-sectional study. Orphanet Journal of Rare Diseases, 2016, 11, 34.	2.7	63
45	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
46	Hard ways towards adulthood: the transition phase in young people with myotonic dystrophy. Acta Myologica, 2016, 35, 145-149.	1.5	2
47	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
48	Muscle exercise in limb girdle muscular dystrophies: pitfall and advantages. Acta Myologica, 2015, 34, 3-8.	1.5	20
49	A multi-parametric protocol to study exercise intolerance in McArdle's disease. Acta Myologica, 2015, 34, 120-125.	1.5	6
50	LMNA -associated myopathies. Neurology, 2014, 83, 1634-1644.	1.1	57
51	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
52	A Mutation in theCASQ1Gene Causes a Vacuolar Myopathy with Accumulation of Sarcoplasmic Reticulum Protein Aggregates. Human Mutation, 2014, 35, 1163-1170.	2.5	53
53	Posterior reversible encephalopathy syndrome in a complicated autoimmune background: differential diagnosis and etiological hypothesis. Acta Neurologica Belgica, 2013, 113, 191-193.	1.1	1
54	An â€œinflammatoryâ€•mitochondrial myopathy. A case report. Neuromuscular Disorders, 2013, 23, 907-910.	0.6	13

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55	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
56	Next-Generation Sequencing Identifies Transportin 3 as the Causative Gene for LGMD1F. PLoS ONE, 2013, 8, e63536.	2.5	69
57	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
58	A personal monitoring architecture to detect muscular fatigue in elderly. Neuromuscular Disorders, 2012, 22, S192-S197.	0.6	11
59	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
60	Nerve, muscle and heart acute toxicity following oxaliplatin and capecitabine treatment. Neuromuscular Disorders, 2012, 22, 767-770.	0.6	7
61	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
62	The empowerment of translational research: lessons from laminopathies. Orphanet Journal of Rare Diseases, 2012, 7, 37.	2.7	7
63	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
64	Effects of aerobic training on exercise-related oxidative stress in mitochondrial myopathies. Neuromuscular Disorders, 2012, 22, S172-S177.	0.6	31
65	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
66	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
67	Inflammatory myopathy in a patient with postural and kinetik tremor. Neurological Sciences, 2011, 32, 1175-1178.	1.9	1
68	Metabolic myopathies: functional evaluation by different exercise testing approaches. Musculoskeletal Surgery, 2011, 95, 59-67.	1.5	15
69	Oxidative Stress Treatment for Clinical Trials in Neurodegenerative Diseases. Journal of Alzheimer's Disease, 2011, 24, 111-126.	2.6	39
70	Pes cavus and hereditary neuropathies: when a relationship should be suspected. Journal of Orthopaedics and Traumatology, 2010, 11, 195-201.	2.3	11
71	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
72	Prevalent cardiac phenotype resulting in heart transplantation in a novel LMNA gene duplication. Neuromuscular Disorders, 2010, 20, 512-516.	0.6	12

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73	A wearable pervasive platform for the intelligent monitoring of muscular fatigue. , 2010, , .		7
74	A pervasive activity management and rehabilitation support system for the elderly. , 2010, , .		3
75	Astrocyteâ€“neuron interactions in neurological disorders. Journal of Biological Physics, 2009, 35, 317-336.	1.5	144
76	An Ontology-Driven Multisensorial Platform to Enable Unobtrusive Human Monitoring and Independent Living. , 2009, , .		8
77	Autosomal dominant psychiatric disorders and mitochondrial DNA multiple deletions: Report of a family. Journal of Affective Disorders, 2008, 106, 173-177.	4.1	27
78	Functional Diagnostics in Mitochondrial Diseases. Bioscience Reports, 2007, 27, 53-67.	2.4	43
79	Mitochondria and Neurodegeneration. Bioscience Reports, 2007, 27, 87-104.	2.4	125
80	Lack of association between mtDNA haplogroups and Alzheimerâ€™s disease in Tuscany. Neurological Sciences, 2007, 28, 142-147.	1.9	41
81	Unusual concomitant smallâ€•and largeâ€fiber neuropathy related to hypereosinophilic syndrome. Clinical and Experimental Neuroimmunology, 0, , .	1.0	0