Giorgio Tasca

List of Publications by Year
in descending order

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125	4,072	87886	144002
papers	citations	38 h-index	57 g-index

129	129	129	5603
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Diagnostic interest of whole-body MRI in early- and late-onset LAMA2 muscular dystrophies: a large international cohort. Journal of Neurology, 2022, 269, 2414-2429.	3.6	5
2	Diagnostic magnetic resonance imaging biomarkers for facioscapulohumeral muscular dystrophy identified by machine learning. Journal of Neurology, 2022, 269, 2055-2063.	3.6	10
3	Upper body involvement in GNE myopathy assessed by muscle imaging. Neuromuscular Disorders, 2022, 32, 410-418.	0.6	2
4	Technology outcome measures in neuromuscular disorders: A systematic review. European Journal of Neurology, 2022, 29, 1266-1278.	3.3	7
5	Muscle-MRI and Functional Levels for the Evaluation of Upper Limbs in Duchenne Muscular Dystrophy: A Critical Review of the Literature. Medicina (Lithuania), 2022, 58, 440.	2.0	1
6	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
7	Dynamic magnetic resonance imaging of muscle contraction in facioscapulohumeral muscular dystrophy. Scientific Reports, 2022, 12, 7250.	3.3	3
8	Texture analysis and machine learning to predict water T2 and fat fraction from non-quantitative MRI of thigh muscles in Facioscapulohumeral muscular dystrophy. European Journal of Radiology, 2021, 134, 109460.	2.6	15
9	Fast Open-Source Toolkit for Water T2 Mapping in the Presence of Fat From Multi-Echo Spin-Echo Acquisitions for Muscle MRI. Frontiers in Neurology, 2021, 12, 630387.	2.4	9
10	INPP5K and SIL1 associated pathologies with overlapping clinical phenotypes converge through dysregulation of PHGDH. Brain, 2021, 144, 2427-2442.	7.6	7
11	Long-term Follow-up and Muscle Imaging Findings in Brachio-Cervical Inflammatory Myopathy. Neurology: Neuroimmunology and NeuroInflammation, 2021, 8, .	6.0	4
12	Anti-cN1A Antibodies Are Associated with More Severe Dysphagia in Sporadic Inclusion Body Myositis. Cells, 2021, 10, 1146.	4.1	23
13	Eyelid closing and opening disorders in patients with unilateral brain lesions: A case report with video neuroimage and a systematic review of the literature. Journal of Clinical Neuroscience, 2021, 87, 69-73.	1.5	3
14	Redox Homeostasis in Muscular Dystrophies. Cells, 2021, 10, 1364.	4.1	16
15	1st FSHD European Trial Network workshop:Working towards trial readiness across Europe. Neuromuscular Disorders, 2021, 31, 907-918.	0.6	9
16	Thr124Met myelin protein zero mutation mimicking motor neuron disease. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2021, , 1-6.	1.7	0
17	High-Throughput Digital Image Analysis Reveals Distinct Patterns of Dystrophin Expression in Dystrophinopathy Patients. Journal of Neuropathology and Experimental Neurology, 2021, 80, 955-965.	1.7	9
18	Proteomics of Muscle Microdialysates Identifies Potential Circulating Biomarkers in Facioscapulohumeral Muscular Dystrophy. International Journal of Molecular Sciences, 2021, 22, 290.	4.1	25

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19	Deep learning for automatic segmentation of thigh and leg muscles. Magnetic Resonance Materials in Physics, Biology, and Medicine, 2021, , 1.	2.0	9
20	Central nervous system immune reconstitution inflammatory syndrome after autologous stem cell transplantation. Bone Marrow Transplantation, 2020, 55, 268-271.	2.4	3
21	European muscle MRI study in limb girdle muscular dystrophy type R1/2A (LGMDR1/LGMD2A). Journal of Neurology, 2020, 267, 45-56.	3.6	43
22	Glycogenin is Dispensable for Glycogen Synthesis in Human Muscle, and Glycogenin Deficiency Causes Polyglucosan Storage. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 557-566.	3.6	17
23	A man with sarcoidosis and slurred speech. European Journal of Neurology, 2020, 27, e7-e8.	3.3	1
24	MRI patterns of muscle involvement in type 2 and 3 spinal muscular atrophy patients. Journal of Neurology, 2020, 267, 898-912.	3.6	32
25	247th ENMC International Workshop: Muscle magnetic resonance imaging - Implementing muscle MRI as a diagnostic tool for rare genetic myopathy cohorts. Hoofddorp, The Netherlands, September 2019. Neuromuscular Disorders, 2020, 30, 938-947.	0.6	11
26	250th ENMC International Workshop: Clinical trial readiness in nemaline myopathy 6â€“8 September 2019, Hoofdorp, the Netherlands. Neuromuscular Disorders, 2020, 30, 866-875.	0.6	6
27	A Recurrent Pathogenic Variant of INPP5K Underlies Autosomal Recessive Congenital Muscular Dystrophy With Cataracts and Intellectual Disability: Evidence for a Founder Effect in Southern Italy. Frontiers in Genetics, 2020, 11, 565868.	2.3	8
28	Genotypeâ€“phenotype correlations in recessive titinopathies. Genetics in Medicine, 2020, 22, 2029-2040.	2.4	35
29	Deep phenotyping of facioscapulohumeral muscular dystrophy type 2 by magnetic resonance imaging. European Journal of Neurology, 2020, 27, 2604-2615.	3.3	16
30	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
31	New genotype-phenotype correlations in a large European cohort of patients with sarcoglycanopathy. Brain, 2020, 143, 2696-2708.	7.6	45
32	Peculiar muscle imaging findings in a patient with alphaB-crystallinopathy and axial myopathy. Journal of the Neurological Sciences, 2020, 416, 116999.	0.6	5
33	Response to: SOD1 mutations in adultâ€onset distal spinal muscular atrophy. European Journal of Neurology, 2020, 27, e74.	3.3	1
34	Accuracy of a machine learning muscle MRI-based tool for the diagnosis of muscular dystrophies. Neurology, 2020, 94, e1094-e1102.	1.1	45
35	SOD1 p.D12Y variant is associated with amyotrophic lateral sclerosis/distal myopathy spectrum. European Journal of Neurology, 2020, 27, 1304-1309.	3.3	4
36	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

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37	Cytidine Diphosphate-Ribitol Analysis for Diagnostics and Treatment Monitoring of Cytidine Diphosphate-l-Ribitol Pyrophosphorylase A Muscular Dystrophy. Clinical Chemistry, 2019, 65, 1295-1306.	3.2	11
38	The variability of SMCHD1 gene in FSHD patients: evidence of new mutations. Human Molecular Genetics, 2019, 28, 3912-3920.	2.9	9
39	Tracking muscle wasting and disease activity in facioscapulohumeral muscular dystrophy by qualitative longitudinal imaging. Journal of Cachexia, Sarcopenia and Muscle, 2019, 10, 1258-1265.	7.3	35
40	MYO-MRI diagnostic protocols in genetic myopathies. Neuromuscular Disorders, 2019, 29, 827-841.	0.6	46
41	Impairments in contractility and cytoskeletal organisation cause nuclear defects in nemaline myopathy. Acta Neuropathologica, 2019, 138, 477-495.	7.7	25
42	An unusual ryanodine receptor 1 (RYR1) phenotype. Neurology, 2019, 92, e1600-e1609.	1.1	16
43	Advances in Quantitative Imaging of Genetic and Acquired Myopathies: Clinical Applications and Perspectives. Frontiers in Neurology, 2019, 10, 78.	2.4	32
44	Muscle hypertrophy in amyloid myopathy. Neuromuscular Disorders, 2019, 29, 150-151.	0.6	3
45	Muscle MRI in a large cohort of patients with oculopharyngeal muscular dystrophy. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 576-585.	1.9	38
46	Novel Homozygous KCNJ10 Mutation in a Patient with Non-syndromic Early-Onset Cerebellar Ataxia. Cerebellum, 2018, 17, 499-503.	2.5	10
47	Interpreting Genetic Variants in Titin in Patients With Muscle Disorders. JAMA Neurology, 2018, 75, 557.	9.0	69
48	Sporadic late-onset nemaline myopathy: clinical, pathology and imaging findings in a single center cohort. Journal of Neurology, 2018, 265, 542-551.	3.6	36
49	Neuromyopathy with congenital cataracts and glaucoma: a distinct syndrome caused by POLG variants. European Journal of Human Genetics, 2018, 26, 367-373.	2.8	3
50	Expanding the histopathological spectrum of CFL2â€related myopathies. Clinical Genetics, 2018, 93, 1234-1239.	2.0	11
51	Muscle Microdialysis to Investigate Inflammatory Biomarkers in Facioscapulohumeral Muscular Dystrophy. Molecular Neurobiology, 2018, 55, 2959-2966.	4.0	25
52	Diagnostic anoctaminâ€5 protein defect in patients with ANO5â€mutated muscular dystrophy. Neuropathology and Applied Neurobiology, 2018, 44, 441-448.	3.2	19
53	MRI in sarcoglycanopathies: a large international cohort study. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 72-77.	1.9	55
54	Hereditary myopathy with early respiratory failure (HMERF): Still rare, but common enough. Neuromuscular Disorders, 2018, 28, 268-276.	0.6	25

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55	Functional levels and MRI patterns of muscle involvement in upper limbs in Duchenne muscular dystrophy. PLoS ONE, 2018, 13, e0199222.	2.5	20
56	The italian limb girdle muscular dystrophy registry: Relative frequency, clinical features, and differential diagnosis. Muscle and Nerve, 2017, 55, 55-68.	2.2	86
57	Rinsing after spinning: plasmapheresis in EBV-related post-infectious cerebellitis. Journal of Neurology, 2017, 264, 576-577.	3.6	3
58	Concentric muscle involvement in POLG -related distal myopathy. Neuromuscular Disorders, 2017, 27, 500-501.	0.6	2
59	Association study reveals novel risk loci for sporadic inclusion body myositis. European Journal of Neurology, 2017, 24, 572-577.	3.3	11
60	1st International Workshop on Clinical trial readiness for sarcoglycanopathies 15â€“16 November 2016, Evry, France. Neuromuscular Disorders, 2017, 27, 683-692.	0.6	9
61	Muscle MRI in neutral lipid storage disease (NLSD). Journal of Neurology, 2017, 264, 1334-1342.	3.6	15
62	Novel mutations in <scp>KARS</scp> cause hypertrophic cardiomyopathy and combined mitochondrial respiratory chain defect. Clinical Genetics, 2017, 91, 918-923.	2.0	27
63	Facilitating orphan drug development: Proceedings of the TREAT-NMD International Conference, December 2015, Washington, DC, USA. Neuromuscular Disorders, 2017, 27, 693-701.	0.6	1
64	Potential therapeutic targets for ALS: MIR206, MIR208b and MIR499 are modulated during disease progression in the skeletal muscle of patients. Scientific Reports, 2017, 7, 9538.	3.3	48
65	Fasciculations in Late-Onset Pompe Disease: A Sign of Motor Neuron Involvement?. Canadian Journal of Neurological Sciences, 2017, 44, 463-464.	0.5	2
66	Somatic mosaicism represents an underestimated event underlying collagen 6-related disorders. European Journal of Paediatric Neurology, 2017, 21, 873-883.	1.6	4
67	Targeted Next-Generation Sequencing Reveals Novel TTN Mutations Causing Recessive Distal Titinopathy. Molecular Neurobiology, 2017, 54, 7212-7223.	4.0	38
68	Matrin 3 variants are frequent in Italian ALS patients. Neurobiology of Aging, 2017, 49, 218.e1-218.e7.	3.1	35
69	Estrogens enhance myoblast differentiation in facioscapulohumeral muscular dystrophy by antagonizing DUX4 activity. Journal of Clinical Investigation, 2017, 127, 1531-1545.	8.2	46
70	MYH7-related myopathies: clinical, histopathological and imaging findings in a cohort of Italian patients. Orphanet Journal of Rare Diseases, 2016, 11, 91.	2.7	70
71	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
72	Start codon mutation of GYG1 causing late-onset polyglucosan body myopathy with nemaline rods. Journal of Neurology, 2016, 263, 2133-2135.	3.6	17

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73	TBCE Mutations Cause Early-Onset Progressive Encephalopathy with Distal Spinal Muscular Atrophy. American Journal of Human Genetics, 2016, 99, 974-983.	6.2	49
74	New ALSâ€Related Genes Expand the Spectrum Paradigm of Amyotrophic Lateral Sclerosis. Brain Pathology, 2016, 26, 266-275.	4.1	26
75	The genetic basis of undiagnosed muscular dystrophies and myopathies. Neurology, 2016, 87, 71-76.	1.1	92
76	Magnetic resonance imaging in a large cohort of facioscapulohumeral muscular dystrophy patients: Pattern refinement and implications for clinical trials. Annals of Neurology, 2016, 79, 854-864.	5.3	83
77	Allele-specific DNA hypomethylation characterises FSHD1 and FSHD2. Journal of Medical Genetics, 2016, 53, 348-355.	3.2	54
78	Clinical trial preparedness in facioscapulohumeral muscular dystrophy: Clinical, tissue, and imaging outcome measures 29â€“30 May 2015, Rochester, New York. Neuromuscular Disorders, 2016, 26, 181-186.	0.6	43
79	A novel AIFM1 mutation expands the phenotype to an infantile motor neuron disease. European Journal of Human Genetics, 2016, 24, 463-466.	2.8	51
80	Muscle imaging in patients with tubular aggregate myopathy caused by mutations in STIM1. Neuromuscular Disorders, 2015, 25, 898-903.	0.6	13
81	Prevalence of congenital muscular dystrophy in Italy. Neurology, 2015, 84, 904-911.	1.1	75
82	Next generation sequencing on patients with LGMD and nonspecific myopathies: Findings associated with ANO5 mutations. Neuromuscular Disorders, 2015, 25, 533-541.	0.6	65
83	Muscle imaging in fibrodysplasia ossificans progressiva: The neurologist's perspective. Neuromuscular Disorders, 2015, 25, 672-673.	0.6	1
84	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
85	Magnetic resonance imaging pattern recognition in sporadic inclusionâ€body myositis. Muscle and Nerve, 2015, 52, 956-962.	2.2	93
86	POPDC1S201F causes muscular dystrophy and arrhythmia by affecting protein trafficking. Journal of Clinical Investigation, 2015, 126, 239-253.	8.2	85
87	Biochemical Characterization of Patients With In-Frame or Out-of-FrameDMDDeletions Pertinent to Exon 44 or 45 Skipping. JAMA Neurology, 2014, 71, 32.	9.0	71
88	Calf muscle involvement in Becker muscular dystrophy: When size does not matter. Journal of the Neurological Sciences, 2014, 347, 301-304.	0.6	16
89	Isolated semitendinosus involvement in the initial stages of limb-girdle muscular dystrophy 2L. Neuromuscular Disorders, 2014, 24, 1118-1119.	0.6	9
90	Childhood onset tubular aggregate myopathy associated with de novo STIM1 mutations. Journal of Neurology, 2014, 261, 870-876.	3.6	56

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91	Hereditary myopathy with early respiratory failure: occurrence in various populations. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 345-353.	1.9	65
92	Clinical, histological and genetic characterisation of patients with tubular aggregate myopathy caused by mutations in STIM1. Journal of Medical Genetics, 2014, 51, 824-833.	3.2	72
93	Upper Girdle Imaging in Facioscapulohumeral Muscular Dystrophy. PLoS ONE, 2014, 9, e100292.	2.5	71
94	Molecular, clinical, and muscle studies in myotonic dystrophy type 1 (DM1) associated with novel variant CCG expansions. Journal of Neurology, 2013, 260, 1245-1257.	3.6	41
95	Somatic mosaicism in TPM2-related myopathy with nemaline rods and cap structures. Acta Neuropathologica, 2013, 125, 169-171.	7.7	15
96	Centronuclear myopathy related to dynamin 2 mutations: Clinical, morphological, muscle imaging and genetic features of an Italian cohort. Neuromuscular Disorders, 2013, 23, 229-238.	0.6	53
97	Myotonic dystrophy type 1 and de novo FSHD mutation double trouble: A clinical and muscle MRI study. Neuromuscular Disorders, 2013, 23, 427-431.	0.6	16
98	Limb-girdle muscular dystrophy with Î±-dystroglycan deficiency and mutations in the ISPD gene. Neurology, 2013, 80, 963-965.	1.1	26
99	Teaching Video Neuro Images : Complicated scapular winging. Neurology, 2013, 81, e95.	1.1	2
100	â€˜Pathognomonicâ€™ muscle imaging findings in <scp>DNAJB</scp>6 mutated <scp>LGMD</scp>1<scp>D</scp>. European Journal of Neurology, 2013, 20, 1553-1559.	3.3	32
101	Mitochondrial Network Genes in the Skeletal Muscle of Amyotrophic Lateral Sclerosis Patients. PLoS ONE, 2013, 8, e57739.	2.5	42
102	Eight new mutations and the expanding phenotype variability in muscular dystrophy caused by ANO5. Neurology, 2012, 78, 897-903.	1.1	99
103	Mutations affecting the cytoplasmic functions of the co-chaperone DNAJB6 cause limb-girdle muscular dystrophy. Nature Genetics, 2012, 44, 450-455.	21.4	226
104	New phenotype and pathology features in MYH7-related distal myopathy. Neuromuscular Disorders, 2012, 22, 640-647.	0.6	41
105	Muscle MRI in Becker muscular dystrophy. Neuromuscular Disorders, 2012, 22, S100-S106.	0.6	67
106	Multi-organ investigation in 16 CADASIL families from central Italy sharing the same R1006C mutation. Neuroscience Letters, 2012, 506, 116-120.	2.1	8
107	Different Molecular Signatures in Magnetic Resonance Imaging-Staged Facioscapulohumeral Muscular Dystrophy Muscles. PLoS ONE, 2012, 7, e38779.	2.5	106
108	Novel FLNC mutation in a patient with myofibrillar myopathy in combination with lateâ€onset cerebellar ataxia. Muscle and Nerve, 2012, 46, 275-282.	2.2	26

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109	Muscle imaging findings in GNE myopathy. Journal of Neurology, 2012, 259, 1358-1365.	3.6	57
110	Muscle <scp>MRI</scp> in female carriers of dystrophinopathy. European Journal of Neurology, 2012, 19, 1256-1260.	3.3	31
111	Mosaic caveolin-3 expression in acquired rippling muscle disease without evidence of myasthenia gravis or acetylcholine receptor autoantibodies. Neuromuscular Disorders, 2011, 21, 194-203.	0.6	16
112	P2.38 Lower limb muscle MRI in a large cohort of FSHD patients. Neuromuscular Disorders, 2011, 21, 671.	0.6	3
113	Abnormal vascular smooth muscle cell proliferation in sural nerve biopsy from a patient with sensorimotor axonal neuropathy. Neuropathology, 2011, 31, 197-198.	1.2	1
114	CD8+ T Cells in Facioscapulohumeral Muscular Dystrophy Patients with Inflammatory Features at Muscle MRI. Journal of Clinical Immunology, 2011, 31, 155-166.	3.8	113
115	Mixed connective tissue disease presenting as a peculiar myositis with poor muscle regeneration. Neurological Sciences, 2011, 32, 171-174.	1.9	4
116	Dystrophin quantification and clinical correlations in Becker muscular dystrophy: implications for clinical trials. Brain, 2011, 134, 3547-3559.	7.6	125
117	An Immunological Analysis of Dystroglycan Subunits: Lessons Learned from a Small Cohort of Non-Congenital Dystrophic Patients. The Open Neurology Journal, 2011, 5, 68-74.	0.4	1
118	Analysis of NCAM helps identify unusual phenotypes of hereditary inclusion-body myopathy. Neurology, 2010, 75, 265-272.	1.1	28
119	A novel HSPB1 mutation in an Italian patient with CMT2/dHMN phenotype. Journal of the Neurological Sciences, 2010, 298, 114-117.	0.6	42
120	A case of CMT 1B due to Val 102/fs null mutation of the MPZ gene presenting as hyperCKemia. Clinical Neurology and Neurosurgery, 2010, 112, 794-797.	1.4	13
121	An Italian case of hereditary myopathy with early respiratory failure (HMERF) not associated with the titin kinase domain R279W mutation. Neuromuscular Disorders, 2010, 20, 730-734.	0.6	15
122	Progressive multifocal leukoencephalopathy in a patient with Franklin disease and hypogammaglobulinemia. Journal of the Neurological Sciences, 2009, 284, 203-204.	0.6	9
123	An Italian family with inclusionâ€body myopathy and frontotemporal dementia due to mutation in the VCP gene. Muscle and Nerve, 2008, 37, 111-114.	2.2	44
124	Gene expression profiling in the early phases of DMD: a constant molecular signature characterizes DMD muscle from early postnatal life throughout disease progression. FASEB Journal, 2007, 21, 1210-1226.	0.5	209
125	Isolation and Characterization of Mesoangioblasts from Facioscapulohumeral Muscular Dystrophy Muscle Biopsies. Stem Cells, 2007, 25, 3173-3182.	3.2	37