Giorgio Tasca

List of Publications by Year
in descending order

125 papers	4,072 citations	87843 38 h-index	143943 57 g-index
129 all docs	129 docs citations	129 times ranked	5603 citing authors

#	Article	IF	Citations
1	Mutations affecting the cytoplasmic functions of the co-chaperone DNAJB6 cause limb-girdle muscular dystrophy. Nature Genetics, 2012, 44, 450-455.	9.4	226
2	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.	0.2	209
3	Dystrophin quantification and clinical correlations in Becker muscular dystrophy: implications for clinical trials. Brain, 2011, 134, 3547-3559.	3.7	125
4	CD8+ T Cells in Facioscapulohumeral Muscular Dystrophy Patients with Inflammatory Features at Muscle MRI. Journal of Clinical Immunology, 2011, 31, 155-166.	2.0	113
5	Different Molecular Signatures in Magnetic Resonance Imaging-Staged Facioscapulohumeral Muscular Dystrophy Muscles. PLoS ONE, 2012, 7, e38779.	1.1	106
6	Eight new mutations and the expanding phenotype variability in muscular dystrophy caused by ANO5. Neurology, 2012, 78, 897-903.	1.5	99
7	Magnetic resonance imaging pattern recognition in sporadic inclusionâ€body myositis. Muscle and Nerve, 2015, 52, 956-962.	1.0	93
8	The genetic basis of undiagnosed muscular dystrophies and myopathies. Neurology, 2016, 87, 71-76.	1.5	92
9	The italian limb girdle muscular dystrophy registry: Relative frequency, clinical features, and differential diagnosis. Muscle and Nerve, 2017, 55, 55-68.	1.0	86
10	POPDC1S201F causes muscular dystrophy and arrhythmia by affecting protein trafficking. Journal of Clinical Investigation, 2015, 126, 239-253.	3.9	85
11	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.	2.8	83
12	Prevalence of congenital muscular dystrophy in Italy. Neurology, 2015, 84, 904-911.	1.5	75
13	Clinical, histological and genetic characterisation of patients with tubular aggregate myopathy caused by mutations in STIM1. Journal of Medical Genetics, 2014, 51, 824-833.	1.5	72
14	Biochemical Characterization of Patients With In-Frame or Out-of-FrameDMDDeletions Pertinent to Exon 44 or 45 Skipping. JAMA Neurology, 2014, 71, 32.	4.5	71
15	Upper Girdle Imaging in Facioscapulohumeral Muscular Dystrophy. PLoS ONE, 2014, 9, e100292.	1.1	71
16	MYH7-related myopathies: clinical, histopathological and imaging findings in a cohort of Italian patients. Orphanet Journal of Rare Diseases, 2016, 11, 91.	1.2	70
17	Interpreting Genetic Variants in Titin in Patients With Muscle Disorders. JAMA Neurology, 2018, 75, 557.	4.5	69
18	Muscle MRI in Becker muscular dystrophy. Neuromuscular Disorders, 2012, 22, S100-S106.	0.3	67

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19	Hereditary myopathy with early respiratory failure: occurrence in various populations. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 345-353.	0.9	65
20	Next generation sequencing on patients with LGMD and nonspecific myopathies: Findings associated with ANO5 mutations. Neuromuscular Disorders, 2015, 25, 533-541.	0.3	65
21	Muscle imaging findings in GNE myopathy. Journal of Neurology, 2012, 259, 1358-1365.	1.8	57
22	Childhood onset tubular aggregate myopathy associated with de novo STIM1 mutations. Journal of Neurology, 2014, 261, 870-876.	1.8	56
23	MRI in sarcoglycanopathies: a large international cohort study. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 72-77.	0.9	55
24	Allele-specific DNA hypomethylation characterises FSHD1 and FSHD2. Journal of Medical Genetics, 2016, 53, 348-355.	1.5	54
25	Centronuclear myopathy related to dynamin 2 mutations: Clinical, morphological, muscle imaging and genetic features of an Italian cohort. Neuromuscular Disorders, 2013, 23, 229-238.	0.3	53
26	Centronuclear myopathies: genotypeâ€“phenotype correlation and frequency of defined genetic forms in an Italian cohort. Journal of Neurology, 2015, 262, 1728-1740.	1.8	51
27	A novel AIFM1 mutation expands the phenotype to an infantile motor neuron disease. European Journal of Human Genetics, 2016, 24, 463-466.	1.4	51
28	TBCE Mutations Cause Early-Onset Progressive Encephalopathy with Distal Spinal Muscular Atrophy. American Journal of Human Genetics, 2016, 99, 974-983.	2.6	49
29	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.	1.6	48
30	MYO-MRI diagnostic protocols in genetic myopathies. Neuromuscular Disorders, 2019, 29, 827-841.	0.3	46
31	Estrogens enhance myoblast differentiation in facioscapulohumeral muscular dystrophy by antagonizing DUX4 activity. Journal of Clinical Investigation, 2017, 127, 1531-1545.	3.9	46
32	New genotype-phenotype correlations in a large European cohort of patients with sarcoglycanopathy. Brain, 2020, 143, 2696-2708.	3.7	45
33	Accuracy of a machine learning muscle MRI-based tool for the diagnosis of muscular dystrophies. Neurology, 2020, 94, e1094-e1102.	1.5	45
34	An Italian family with inclusionâ€body myopathy and frontotemporal dementia due to mutation in the VCP gene. Muscle and Nerve, 2008, 37, 111-114.	1.0	44
35	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.	0.3	43
36	European muscle MRI study in limb girdle muscular dystrophy type R1/2A (LGMDR1/LGMD2A). Journal of Neurology, 2020, 267, 45-56.	1.8	43

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37	A novel HSPB1 mutation in an Italian patient with CMT2/dHMN phenotype. Journal of the Neurological Sciences, 2010, 298, 114-117.	0.3	42
38	Mitochondrial Network Genes in the Skeletal Muscle of Amyotrophic Lateral Sclerosis Patients. PLoS ONE, 2013, 8, e57739.	1.1	42
39	New phenotype and pathology features in MYH7-related distal myopathy. Neuromuscular Disorders, 2012, 22, 640-647.	0.3	41
40	Molecular, clinical, and muscle studies in myotonic dystrophy type 1 (DM1) associated with novel variant CCG expansions. Journal of Neurology, 2013, 260, 1245-1257.	1.8	41
41	Targeted Next-Generation Sequencing Reveals Novel TTN Mutations Causing Recessive Distal Titinopathy. Molecular Neurobiology, 2017, 54, 7212-7223.	1.9	38
42	Muscle MRI in a large cohort of patients with oculopharyngeal muscular dystrophy. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 576-585.	0.9	38
43	Isolation and Characterization of Mesoangioblasts from Facioscapulohumeral Muscular Dystrophy Muscle Biopsies. Stem Cells, 2007, 25, 3173-3182.	1.4	37
44	Sporadic late-onset nemaline myopathy: clinical, pathology and imaging findings in a single center cohort. Journal of Neurology, 2018, 265, 542-551.	1.8	36
45	Matrin 3 variants are frequent in Italian ALS patients. Neurobiology of Aging, 2017, 49, 218.e1-218.e7.	1.5	35
46	Tracking muscle wasting and disease activity in facioscapulohumeral muscular dystrophy by qualitative longitudinal imaging. Journal of Cachexia, Sarcopenia and Muscle, 2019, 10, 1258-1265.	2.9	35
47	Genotypeâ€“phenotype correlations in recessive titinopathies. Genetics in Medicine, 2020, 22, 2029-2040.	1.1	35
48	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.	1.7	32
49	Advances in Quantitative Imaging of Genetic and Acquired Myopathies: Clinical Applications and Perspectives. Frontiers in Neurology, 2019, 10, 78.	1.1	32
50	MRI patterns of muscle involvement in type 2 and 3 spinal muscular atrophy patients. Journal of Neurology, 2020, 267, 898-912.	1.8	32
51	Muscle <scp>MRI</scp> in female carriers of dystrophinopathy. European Journal of Neurology, 2012, 19, 1256-1260.	1.7	31
52	Analysis of NCAM helps identify unusual phenotypes of hereditary inclusion-body myopathy. Neurology, 2010, 75, 265-272.	1.5	28
53	Novel mutations in <scp>KARS</scp> cause hypertrophic cardiomyopathy and combined mitochondrial respiratory chain defect. Clinical Genetics, 2017, 91, 918-923.	1.0	27
54	Novel FLNC mutation in a patient with myofibrillar myopathy in combination with lateâ€onset cerebellar ataxia. Muscle and Nerve, 2012, 46, 275-282.	1.0	26

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55	Limb-girdle muscular dystrophy with Î±-dystroglycan deficiency and mutations in the ISPD gene. Neurology, 2013, 80, 963-965.	1.5	26
56	New ALSâ€Related Genes Expand the Spectrum Paradigm of Amyotrophic Lateral Sclerosis. Brain Pathology, 2016, 26, 266-275.	2.1	26
57	Muscle Microdialysis to Investigate Inflammatory Biomarkers in Facioscapulohumeral Muscular Dystrophy. Molecular Neurobiology, 2018, 55, 2959-2966.	1.9	25
58	Hereditary myopathy with early respiratory failure (HMERF): Still rare, but common enough. Neuromuscular Disorders, 2018, 28, 268-276.	0.3	25
59	Impairments in contractility and cytoskeletal organisation cause nuclear defects in nemaline myopathy. Acta Neuropathologica, 2019, 138, 477-495.	3.9	25
60	Proteomics of Muscle Microdialysates Identifies Potential Circulating Biomarkers in Facioscapulohumeral Muscular Dystrophy. International Journal of Molecular Sciences, 2021, 22, 290.	1.8	25
61	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
62	Anti-cN1A Antibodies Are Associated with More Severe Dysphagia in Sporadic Inclusion Body Myositis. Cells, 2021, 10, 1146.	1.8	23
63	Functional levels and MRI patterns of muscle involvement in upper limbs in Duchenne muscular dystrophy. PLoS ONE, 2018, 13, e0199222.	1.1	20
64	Diagnostic anoctaminâ€5 protein defect in patients with ANO5â€mutated muscular dystrophy. Neuropathology and Applied Neurobiology, 2018, 44, 441-448.	1.8	19
65	New Mutations in NEB Gene Discovered by Targeted Next-Generation Sequencing in Nemaline Myopathy Italian Patients. Journal of Molecular Neuroscience, 2016, 59, 351-359.	1.1	17
66	Start codon mutation of GYG1 causing late-onset polyglucosan body myopathy with nemaline rods. Journal of Neurology, 2016, 263, 2133-2135.	1.8	17
67	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.	1.8	17
68	Mosaic caveolin-3 expression in acquired rippling muscle disease without evidence of myasthenia gravis or acetylcholine receptor autoantibodies. Neuromuscular Disorders, 2011, 21, 194-203.	0.3	16
69	Myotonic dystrophy type 1 and de novo FSHD mutation double trouble: A clinical and muscle MRI study. Neuromuscular Disorders, 2013, 23, 427-431.	0.3	16
70	Calf muscle involvement in Becker muscular dystrophy: When size does not matter. Journal of the Neurological Sciences, 2014, 347, 301-304.	0.3	16
71	An unusual ryanodine receptor 1 (RYR1) phenotype. Neurology, 2019, 92, e1600-e1609.	1.5	16
72	Deep phenotyping of facioscapulohumeral muscular dystrophy type 2 by magnetic resonance imaging. European Journal of Neurology, 2020, 27, 2604-2615.	1.7	16

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73	Redox Homeostasis in Muscular Dystrophies. Cells, 2021, 10, 1364.	1.8	16
74	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.	0.3	15
75	Somatic mosaicism in TPM2-related myopathy with nemaline rods and cap structures. Acta Neuropathologica, 2013, 125, 169-171.	3.9	15
76	Muscle MRI in neutral lipid storage disease (NLSD). Journal of Neurology, 2017, 264, 1334-1342.	1.8	15
77	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.	1.2	15
78	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.	0.6	13
79	Muscle imaging in patients with tubular aggregate myopathy caused by mutations in STIM1. Neuromuscular Disorders, 2015, 25, 898-903.	0.3	13
80	Association study reveals novel risk loci for sporadic inclusion body myositis. European Journal of Neurology, 2017, 24, 572-577.	1.7	11
81	Expanding the histopathological spectrum of CFL2â€related myopathies. Clinical Genetics, 2018, 93, 1234-1239.	1.0	11
82	Cytidine Diphosphate-Ribitol Analysis for Diagnostics and Treatment Monitoring of Cytidine Diphosphate-l-Ribitol Pyrophosphorylase A Muscular Dystrophy. Clinical Chemistry, 2019, 65, 1295-1306.	1.5	11
83	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.	0.3	11
84	Novel Homozygous KCNJ10 Mutation in a Patient with Non-syndromic Early-Onset Cerebellar Ataxia. Cerebellum, 2018, 17, 499-503.	1.4	10
85	Diagnostic magnetic resonance imaging biomarkers for facioscapulohumeral muscular dystrophy identified by machine learning. Journal of Neurology, 2022, 269, 2055-2063.	1.8	10
86	Progressive multifocal leukoencephalopathy in a patient with Franklin disease and hypogammaglobulinemia. Journal of the Neurological Sciences, 2009, 284, 203-204.	0.3	9
87	Isolated semitendinosus involvement in the initial stages of limb-girdle muscular dystrophy 2L. Neuromuscular Disorders, 2014, 24, 1118-1119.	0.3	9
88	1st International Workshop on Clinical trial readiness for sarcoglycanopathies 15â€“16 November 2016, Evry, France. Neuromuscular Disorders, 2017, 27, 683-692.	0.3	9
89	The variability of SMCHD1 gene in FSHD patients: evidence of new mutations. Human Molecular Genetics, 2019, 28, 3912-3920.	1.4	9
90	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.	1.1	9

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91	1st FSHD European Trial Network workshop:Working towards trial readiness across Europe. Neuromuscular Disorders, 2021, 31, 907-918.	0.3	9
92	High-Throughput Digital Image Analysis Reveals Distinct Patterns of Dystrophin Expression in Dystrophinopathy Patients. Journal of Neuropathology and Experimental Neurology, 2021, 80, 955-965.	0.9	9
93	Deep learning for automatic segmentation of thigh and leg muscles. Magnetic Resonance Materials in Physics, Biology, and Medicine, 2021, , 1.	1.1	9
94	Multi-organ investigation in 16 CADASIL families from central Italy sharing the same R1006C mutation. Neuroscience Letters, 2012, 506, 116-120.	1.0	8
95	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.	1.1	8
96	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.	1.1	7
97	INPP5K and SIL1 associated pathologies with overlapping clinical phenotypes converge through dysregulation of PHGDH. Brain, 2021, 144, 2427-2442.	3.7	7
98	Technology outcome measures in neuromuscular disorders: A systematic review. European Journal of Neurology, 2022, 29, 1266-1278.	1.7	7
99	250th ENMC International Workshop: Clinical trial readiness in nemaline myopathy 6â€“8 September 2019, Hoofdorp, the Netherlands. Neuromuscular Disorders, 2020, 30, 866-875.	0.3	6
100	Peculiar muscle imaging findings in a patient with alphaB-crystallinopathy and axial myopathy. Journal of the Neurological Sciences, 2020, 416, 116999.	0.3	5
101	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.	1.8	5
102	Mixed connective tissue disease presenting as a peculiar myositis with poor muscle regeneration. Neurological Sciences, 2011, 32, 171-174.	0.9	4
103	Somatic mosaicism represents an underestimated event underlying collagen 6-related disorders. European Journal of Paediatric Neurology, 2017, 21, 873-883.	0.7	4
104	SOD1 p.D12Y variant is associated with amyotrophic lateral sclerosis/distal myopathy spectrum. European Journal of Neurology, 2020, 27, 1304-1309.	1.7	4
105	Long-term Follow-up and Muscle Imaging Findings in Brachio-Cervical Inflammatory Myopathy. Neurology: Neuroimmunology and NeuroInflammation, 2021, 8, .	3.1	4
106	P2.38 Lower limb muscle MRI in a large cohort of FSHD patients. Neuromuscular Disorders, 2011, 21, 671.	0.3	3
107	Rinsing after spinning: plasmapheresis in EBV-related post-infectious cerebellitis. Journal of Neurology, 2017, 264, 576-577.	1.8	3
108	Neuromyopathy with congenital cataracts and glaucoma: a distinct syndrome caused by POLG variants. European Journal of Human Genetics, 2018, 26, 367-373.	1.4	3

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109	Muscle hypertrophy in amyloid myopathy. Neuromuscular Disorders, 2019, 29, 150-151.	0.3	3
110	Central nervous system immune reconstitution inflammatory syndrome after autologous stem cell transplantation. Bone Marrow Transplantation, 2020, 55, 268-271.	1.3	3
111	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.	0.8	3
112	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.	2.4	3
113	Dynamic magnetic resonance imaging of muscle contraction in facioscapulohumeral muscular dystrophy. Scientific Reports, 2022, 12, 7250.	1.6	3
114	Teaching Video Neuro Images : Complicated scapular winging. Neurology, 2013, 81, e95.	1.5	2
115	Concentric muscle involvement in POLG -related distal myopathy. Neuromuscular Disorders, 2017, 27, 500-501.	0.3	2
116	Fasciculations in Late-Onset Pompe Disease: A Sign of Motor Neuron Involvement?. Canadian Journal of Neurological Sciences, 2017, 44, 463-464.	0.3	2
117	Upper body involvement in GNE myopathy assessed by muscle imaging. Neuromuscular Disorders, 2022, 32, 410-418.	0.3	2
118	Abnormal vascular smooth muscle cell proliferation in sural nerve biopsy from a patient with sensorimotor axonal neuropathy. Neuropathology, 2011, 31, 197-198.	0.7	1
119	Muscle imaging in fibrodysplasia ossificans progressiva: The neurologist's perspective. Neuromuscular Disorders, 2015, 25, 672-673.	0.3	1
120	Facilitating orphan drug development: Proceedings of the TREAT-NMD International Conference, December 2015, Washington, DC, USA. Neuromuscular Disorders, 2017, 27, 693-701.	0.3	1
121	A man with sarcoidosis and slurred speech. European Journal of Neurology, 2020, 27, e7-e8.	1.7	1
122	Response to: SOD1 mutations in adultâ€onset distal spinal muscular atrophy. European Journal of Neurology, 2020, 27, e74.	1.7	1
123	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
124	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.	0.8	1
125	Thr124Met myelin protein zero mutation mimicking motor neuron disease. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2021, , 1-6.	1.1	0