Stefano Carlo Previtali

List of Publications by Year
in descending order

133 papers	5,886 citations	61857 43 h-index	85405 71 g-index
138 all docs	138 docs citations	138 times ranked	8877 citing authors

#	Article	IF	Citations
1	JAB1 deletion in oligodendrocytes causes senescence-induced inflammation and neurodegeneration in mice. Journal of Clinical Investigation, 2022, 132, .	3.9	12
2	Dipeptidyl peptidase 4/CD26 expression in human idiopathic inflammatory myopathies reveals skeletal muscle injury and vascular inflammation. Clinical and Experimental Rheumatology, 2022, 40, 237-246.	0.4	0
3	Phenotypic Spectrum of Dystrophinopathy Due to Duchenne Muscular Dystrophy Exon 2 Duplications. Neurology, 2022, 98, .	1.5	6
4	Genetic modifiers of upper limb function in Duchenne muscular dystrophy. Journal of Neurology, 2022, 269, 4884-4894.	1.8	2
5	Alteration of the late endocytic pathway in Charcotâ€“Marieâ€“Tooth type 2B disease. Cellular and Molecular Life Sciences, 2021, 78, 351-372.	2.4	27
6	A 5-year clinical follow-up study from the Italian National Registry for FSHD. Journal of Neurology, 2021, 268, 356-366.	1.8	15
7	Begelomab for severe refractory dermatomyositis. Medicine (United States), 2021, 100, e24372.	0.4	1
8	Loss of function <scp>MPZ</scp> mutation causes milder <scp>CMT1B</scp> neuropathy. Journal of the Peripheral Nervous System, 2021, 26, 177-183.	1.4	15
9	Rebalancing expression of HMGB1 redox isoforms to counteract muscular dystrophy. Science Translational Medicine, 2021, 13, .	5.8	26
10	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.	1.1	6
11	The nonsense mutation stop+4 model correlates with motor changes in Duchenne muscular dystrophy. Neuromuscular Disorders, 2021, 31, 479-488.	0.3	0
12	Peripheral Nerve Development and the Pathogenesis of Peripheral Neuropathy: the Sorting Point. Neurotherapeutics, 2021, 18, 2156-2168.	2.1	9
13	Merosin deficient congenital muscular dystrophy type 1A: An international workshop on the road to therapy 15-17 November 2019, Maastricht, the Netherlands. Neuromuscular Disorders, 2021, 31, 673-680.	0.3	2
14	Myogenic Cell Transplantation in Genetic and Acquired Diseases of Skeletal Muscle. Frontiers in Genetics, 2021, 12, 702547.	1.1	18
15	Out-of-Frame Mutations in ACTN2 Last Exon Cause a Dominant Distal Myopathy With Facial Weakness. Neurology: Genetics, 2021, 7, e619.	0.9	7
16	Animal Models as a Tool to Design Therapeutical Strategies for CMT-like Hereditary Neuropathies. Brain Sciences, 2021, 11, 1237.	1.1	4
17	Editorial: Current Insights Into LAMA2 Disease. Frontiers in Molecular Neuroscience, 2021, 14, 780635.	1.4	1
18	Dipeptidyl peptidase 4/CD26 expression in human idiopathic inflammatory myopathies reveals skeletal muscle injury and vascular inflammation. Clinical and Experimental Rheumatology, 2021, , .	0.4	0

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19	Prostaglandin D2 synthase modulates macrophage activity and accumulation in injured peripheral nerves. Glia, 2020, 68, 95-110.	2.5	13
20	Impaired turnover of hyperfused mitochondria in severe axonal neuropathy due to a novel DRP1 mutation. Human Molecular Genetics, 2020, 29, 177-188.	1.4	30
21	Neuromyelitis optica and myotonic dystrophy type 2: a rare association with diagnostic implications. Journal of Neurology, 2020, 267, 2744-2746.	1.8	4
22	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.	0.9	99
23	Large genotypeâ€“phenotype study in carriers of D4Z4 borderline alleles provides guidance for facioscapulohumeral muscular dystrophy diagnosis. Scientific Reports, 2020, 10, 21648.	1.6	16
24	Phenotypic Variability Among Patients With D4Z4 Reduced Allele Facioscapulohumeral Muscular Dystrophy. JAMA Network Open, 2020, 3, e204040.	2.8	25
25	Rimeporide as a ï¬rst- in-class NHE-1 inhibitor: Results of a phase Ib trial in young patients with Duchenne Muscular Dystrophy. Pharmacological Research, 2020, 159, 104999.	3.1	24
26	Genetic modifiers of respiratory function in Duchenne muscular dystrophy. Annals of Clinical and Translational Neurology, 2020, 7, 786-798.	1.7	36
27	Sodium Channel Myotonia Due to Novel Mutations in Domain I of Nav1.4. Frontiers in Neurology, 2020, 11, 255.	1.1	5
28	LAMA2 Neuropathies: Human Findings and Pathomechanisms From Mouse Models. Frontiers in Molecular Neuroscience, 2020, 13, 60.	1.4	18
29	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
30	P.151Motor performances in exon-2 duplication of the dystrophin gene. Neuromuscular Disorders, 2019, 29, S92-S93.	0.3	0
31	P.267Modifiers of respiratory and cardiac function in the Italian Duchenne muscular dystrophy network and CINRG Duchenne natural history study. Neuromuscular Disorders, 2019, 29, S145.	0.3	1
32	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.	1.1	47
33	Expanding the spectrum of genes responsible for hereditary motor neuropathies. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 1171-1179.	0.9	30
34	A multicenter retrospective study of charcotâ€marieâ€tooth disease type 4B (CMT4B) associated with mutations in myotubularinâ€related proteins (MTMRs). Annals of Neurology, 2019, 86, 55-67.	2.8	35
35	Expanding the central nervous system disease spectrum associated with FLNC mutation. Muscle and Nerve, 2019, 59, E33-E37.	1.0	3
36	Cardiac and Neuromuscular Features of Patients WithLMNA-Related Cardiomyopathy. Annals of Internal Medicine, 2019, 171, 458.	2.0	33

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37	AB1136â€…CD26: A POTENTIAL NOVEL HISTOLOGICAL MARKER OF IDIOPATHIC INFLAMMATORY MYOPATHIES. , 2019, , .		0
38	Effects of shortâ€toâ€long term enzyme replacement therapy (ERT) on skeletal muscle tissue in late onset Pompe disease (LOPD). Neuropathology and Applied Neurobiology, 2018, 44, 449-462.	1.8	23
39	Vocal cord paralysis in Charcotâ€“Marieâ€“Tooth type 4b1 disease associated with a novel mutation in the myotubularin-related protein 2 gene: A case report and review of the literature. Neuromuscular Disorders, 2017, 27, 487-491.	0.3	26
40	Diagnosis of Duchenne Muscular Dystrophy in Italy in the last decade: Critical issues and areas for improvements. Neuromuscular Disorders, 2017, 27, 447-451.	0.3	42
41	Jab1 in the pathogenesis of Merosin deficient congenital muscular dystrophy (MDC1A). Neuromuscular Disorders, 2017, 27, S108.	0.3	1
42	Rimeporide: safety, tolerability and pharmacokinetic results from a phase Ib study in DMD boys as well as exploratory biomarkers. Neuromuscular Disorders, 2017, 27, S215-S216.	0.3	3
43	Kif13b Regulates PNS and CNS Myelination through the Dlg1 Scaffold. PLoS Biology, 2016, 14, e1002440.	2.6	32
44	Timed Rise from Floor as a Predictor of Disease Progression in Duchenne Muscular Dystrophy: An Observational Study. PLoS ONE, 2016, 11, e0151445.	1.1	32
45	Niacinâ€mediated Tace activation ameliorates <scp>CMT</scp> neuropathies with focal hypermyelination. EMBO Molecular Medicine, 2016, 8, 1438-1454.	3.3	48
46	Registries versus tertiary care centers: How do we measure standards of care in Duchenne muscular dystrophy?. Neuromuscular Disorders, 2016, 26, 261-263.	0.3	3
47	Longitudinal <scp>MRI</scp> quantification of muscle degeneration in Duchenne muscular dystrophy. Annals of Clinical and Translational Neurology, 2016, 3, 607-622.	1.7	50
48	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.	0.3	57
49	How Schwann Cells Sort Axons. Neuroscientist, 2016, 22, 252-265.	2.6	147
50	Muscle MRI findings in facioscapulohumeral muscular dystrophy. European Radiology, 2016, 26, 693-705.	2.3	48
51	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.	0.9	17
52	Intraâ€arterial transplantation of <scp>HLA</scp> â€matched donor mesoangioblasts in Duchenne muscular dystrophy. EMBO Molecular Medicine, 2015, 7, 1513-1528.	3.3	146
53	ISPD mutations account for a small proportion of Italian Limb Girdle Muscular Dystrophy cases. BMC Neurology, 2015, 15, 172.	0.8	10
54	Mesoangioblast delivery of miniagrin ameliorates murine model of merosin-deficient congenital muscular dystrophy type 1A. Skeletal Muscle, 2015, 5, 30.	1.9	15

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55	A novel heat shock protein 27 homozygous mutation: widening of the continuum between MND/dHMN/CMT2. Journal of the Peripheral Nervous System, 2015, 20, 419-421.	1.4	11
56	Novel Splice-Site Mutation in SMN1 Associated with a very Severe SMA-I Phenotype. Journal of Molecular Neuroscience, 2015, 56, 212-215.	1.1	11
57	A new double-trouble phenotype: fascioscapulohumeral muscular dystrophy ameliorates hereditary spastic paraparesis due to spastin mutation. Journal of Neurology, 2015, 262, 476-478.	1.8	8
58	The expanding spectrum of PRPS1-associated phenotypes: three novel mutations segregating with X-linked hearing loss and mild peripheral neuropathy. European Journal of Human Genetics, 2015, 23, 766-773.	1.4	22
59	Loss of Fig4 in both Schwann cells and motor neurons contributes to CMT4J neuropathy. Human Molecular Genetics, 2015, 24, 383-396.	1.4	39
60	Combined cell and gene therapy to treat merosin deficient congenital muscular dystrophy. Neuromuscular Disorders, 2015, 25, S270.	0.3	0
61	Clinical and molecular features of a large cohort of Italian McArdle patients. Neuromuscular Disorders, 2015, 25, S219.	0.3	0
62	miRNAs as serum biomarkers for Duchenne muscular dystrophy: Correlation analysis in a multicentre study between miRNA levels and clinical status of DMD patients. Neuromuscular Disorders, 2015, 25, S252.	0.3	0
63	Long Term Natural History Data in Ambulant Boys with Duchenne Muscular Dystrophy: 36-Month Changes. PLoS ONE, 2014, 9, e108205.	1.1	98
64	LMNA -associated myopathies. Neurology, 2014, 83, 1634-1644.	1.5	57
65	Brain connectivity abnormalities extend beyond the sensorimotor network in peripheral neuropathy. Human Brain Mapping, 2014, 35, 513-526.	1.9	15
66	Jab1 regulates Schwann cell proliferation and axonal sorting through p27. Journal of Experimental Medicine, 2014, 211, 29-43.	4.2	35
67	6 Minute Walk Test in Duchenne MD Patients with Different Mutations: 12 Month Changes. PLoS ONE, 2014, 9, e83400.	1.1	65
68	Increased expression of Myosin binding protein H in the skeletal muscle of amyotrophic lateral sclerosis patients. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2014, 1842, 99-106.	1.8	49
69	Autocrine and immune cell-derived BDNF in human skeletal muscle: implications for myogenesis and tissue regeneration. Journal of Pathology, 2013, 231, 190-198.	2.1	40
70	Evaluation of muscle biopsy in late-onset GSDII patients before and after enzyme replacement therapy (ERT). BMC Musculoskeletal Disorders, 2013, 14, P13.	0.8	0
71	P.2.7 6min walk test 12month changes in DMD: Correlation with genotype. Neuromuscular Disorders, 2013, 23, 750-751.	0.3	1
72	24 Month Longitudinal Data in Ambulant Boys with Duchenne Muscular Dystrophy. PLoS ONE, 2013, 8, e52512.	1.1	99

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73	Jab1 regulates Schwann cell proliferation and axonal sorting through p27. Journal of Cell Biology, 2013, 203, 2036OIA155.	2.3	0
74	Loss-of-function mutations in HINT1 cause axonal neuropathy with neuromyotonia. Nature Genetics, 2012, 44, 1080-1083.	9.4	102
75	Importance of SPP1 genotype as a covariate in clinical trials in Duchenne muscular dystrophy. Neurology, 2012, 79, 159-162.	1.5	81
76	Protein profiling reveals energy metabolism and cytoskeletal protein alterations in LMNA mutation carriers. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 970-979.	1.8	16
77	Quantitative muscle strength assessment in duchenne muscular dystrophy: longitudinal study and correlation with functional measures. BMC Neurology, 2012, 12, 91.	0.8	52
78	The empowerment of translational research: lessons from laminopathies. Orphanet Journal of Rare Diseases, 2012, 7, 37.	1.2	7
79	Vimentin regulates peripheral nerve myelination. Development (Cambridge), 2012, 139, 1359-1367.	1.2	58
80	Urokinase Plasminogen Receptor and the Fibrinolytic Complex Play a Role in Nerve Repair after Nerve Crush in Mice, and in Human Neuropathies. PLoS ONE, 2012, 7, e32059.	1.1	16
81	Vimentin regulates peripheral nerve myelination. Journal of Cell Science, 2012, 125, e1-e1.	1.2	0
82	POEMS syndrome. Current Opinion in Neurology, 2011, 24, 491-496.	1.8	68
83	Motor nerve biopsy: Clinical usefulness and histopathological criteria. Annals of Neurology, 2011, 69, 197-201.	2.8	38
84	Functional changes in Duchenne muscular dystrophy. Neurology, 2011, 77, 250-256.	1.5	151
85	Foot pad skin biopsy in mouse models of hereditary neuropathy. Glia, 2010, 58, 2005-2016.	2.5	13
86	Analyzing Histopathological Features of Rare Charcot-Marie-Tooth Neuropathies to Unravel Their Pathogenesis. Archives of Neurology, 2010, 67, 1498-505.	4.9	48
87	P4.35 Outcome measures validation study for mesoangioblasts transplantation in children affected by Duchenne muscular dystrophy. Neuromuscular Disorders, 2010, 20, 668-669.	0.3	0
88	Dlg1, Sec8, and Mtmr2 Regulate Membrane Homeostasis in Schwann Cell Myelination. Journal of Neuroscience, 2009, 29, 8858-8870.	1.7	101
89	Matrix metalloproteinaseâ€2 is involved in myelination of dorsal root ganglia neurons. Glia, 2009, 57, 479-489.	2.5	50
90	Diffuse intraneural leiomyoma in a case of sensorimotor neuropathy. Acta Neuropathologica, 2009, 117, 595-597.	3.9	3

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91	Novel Twinkle (PEO1) gene mutations in mendelian progressive external ophthalmoplegia. Journal of Neurology, 2008, 255, 1384-1391.	1.8	45
92	Inclusion body myopathy, Pagetâ€™s disease of the bone and frontotemporal dementia: recurrence of the VCP R155H mutation in an Italian family and implications for genetic counselling. Clinical Genetics, 2008, 74, 54-60.	1.0	37
93	The extracellular matrix affects axonal regeneration in peripheral neuropathies. Neurology, 2008, 71, 322-331.	1.5	32
94	LYMPHOMATOUS NEUROPATHY IN COLD AGGLUTININ DISEASE. Neurology, 2008, 70, 1715-1716.	1.5	3
95	The Mitochondrial Protease AFG3L2 Is Essential for Axonal Development. Journal of Neuroscience, 2008, 28, 2827-2836.	1.7	92
96	Â6Â4 Integrin and Dystroglycan Cooperate to Stabilize the Myelin Sheath. Journal of Neuroscience, 2008, 28, 6714-6719.	1.7	78
97	Charcotâ€“Marieâ€“Tooth type 4B demyelinating neuropathy: deciphering the role of MTMR phosphatases. Expert Reviews in Molecular Medicine, 2007, 9, 1-16.	1.6	62
98	Phenotypic clustering of lamin A/C mutations in neuromuscular patients. Neurology, 2007, 69, 1285-1292.	1.5	120
99	Hypokalemic periodic paralysis in a patient with acquired growth hormone deficiency. Journal of Endocrinological Investigation, 2007, 30, 341-345.	1.8	2
100	Desmin accumulation restrictive cardiomyopathy and atrioventricular block associated with desmin gene defects. European Journal of Heart Failure, 2006, 8, 477-483.	2.9	153
101	Loss of glial fibrillary acidic protein (GFAP) impairs Schwann cell proliferation and delays nerve regeneration after damage. Journal of Cell Science, 2006, 119, 3981-3993.	1.2	174
102	Different Intracellular Pathomechanisms Produce Diverse Myelin Protein Zero Neuropathies in Transgenic Mice. Journal of Neuroscience, 2006, 26, 2358-2368.	1.7	144
103	Dominant LMNA mutations can cause combined muscular dystrophy and peripheral neuropathy. Journal of Neurology, Neurosurgery and Psychiatry, 2005, 76, 1019-1021.	0.9	67
104	Polyneuropathy in POEMS syndrome: role of angiogenic factors in the pathogenesis. Brain, 2005, 128, 1911-1920.	3.7	216
105	Loss of Mtmr2 Phosphatase in Schwann Cells But Not in Motor Neurons Causes Charcot-Marie-Tooth Type 4B1 Neuropathy with Myelin Outfoldings. Journal of Neuroscience, 2005, 25, 8567-8577.	1.7	95
106	Schwann cell overexpression of the GPR7 receptor in inflammatory and painful neuropathies. Molecular and Cellular Neurosciences, 2005, 28, 55-63.	1.0	23
107	Disruption of Mtmr2 produces CMT4B1-like neuropathy with myelin outfolding and impaired spermatogenesis. Journal of Cell Biology, 2004, 167, 711-721.	2.3	167
108	Schwann cells synthesize Î±7Î²1 integrin which is dispensable for peripheral nerve development and myelination. Molecular and Cellular Neurosciences, 2003, 23, 210-218.	1.0	44

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109	Hypogonadotropic hypogonadism and peripheral neuropathy inEbf2-null mice. Development (Cambridge), 2003, 130, 401-410.	1.2	89
110	Myotubularin-related 2 protein phosphatase and neurofilament light chain protein, both mutated in CMT neuropathies, interact in peripheral nerve. Human Molecular Genetics, 2003, 12, 1713-1723.	1.4	67
111	Expression of Laminin Receptors in Schwann Cell Differentiation: Evidence for Distinct Roles. Journal of Neuroscience, 2003, 23, 5520-5530.	1.7	100
112	Autoimmunity in the Peripheral Nervous System. Critical Reviews in Neurobiology, 2003, 15, 1-39.	3.3	15
113	Autoantibodies to Amphiphysin I and Amphiphysin II in a Patient with Sensory-Motor Neuropathy. European Neurology, 2002, 47, 196-200.	0.6	13
114	Conditional disruption of Î²1 integrin in Schwann cells impedes interactions with axons. Journal of Cell Biology, 2002, 156, 199-210.	2.3	294
115	Changes in expression of the orphan G-protein coupled receptor GPR7 in human painful peripheral neuropathies. Pharmacochemistry Library, 2002, , 115-124.	0.1	0
116	Role of integrins in the peripheral nervous system. Progress in Neurobiology, 2001, 64, 35-49.	2.8	123
117	Human IgM anti-GM1 autoantibodies modulate intracellular calcium homeostasis in neuroblastoma cells. Journal of Neuroimmunology, 2001, 114, 213-219.	1.1	31
118	Expression of Angiopoietin-1 in Human Glioblastomas Regulates Tumor-Induced Angiogenesis. Arteriosclerosis, Thrombosis, and Vascular Biology, 2001, 21, 536-541.	1.1	50
119	P0 Glycoprotein Overexpression Causes Congenital Hypomyelination of Peripheral Nerves. Journal of Cell Biology, 2000, 148, 1021-1034.	2.3	145
120	Epitope-Tagged P0Glycoprotein Causes Charcot-Marie-Toothâ€“Like Neuropathy in Transgenic Mice. Journal of Cell Biology, 2000, 151, 1035-1046.	2.3	53
121	A novel P0glycoprotein transgene activates expression oflacZ in myelin-forming Schwann cells. European Journal of Neuroscience, 1999, 11, 1577-1586.	1.2	57
122	P0-CreTransgenic Mice for Inactivation of Adhesion Molecules in Schwann Cells. Annals of the New York Academy of Sciences, 1999, 883, 116-123.	1.8	179
123	Peripheral Nerve Dysmyelination Due to P0Glycoprotein Overexpression Is Dose-Dependent. Annals of the New York Academy of Sciences, 1999, 883, 294-301.	1.8	5
124	Docetaxel neuropathy: a distal axonopathy. Acta Neuropathologica, 1999, 98, 651-653.	3.9	35
125	Laminin receptor ?6?4 integrin is highly expressed in ENU-induced glioma in rat. Glia, 1999, 26, 55-63.	2.5	7
126	The role of integrins in immune-mediated diseases of the nervous system. Trends in Neurosciences, 1999, 22, 30-38.	4.2	128

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127	Laminin receptor alpha6beta4 integrin is highly expressed in ENU-induced glioma in rat. Glia, 1999, 26, 55-63.	2.5	3
128	Isolation and characterization of an oligodendrocyte precursor-derived B-cell epitope in multiple sclerosis. Annals of Neurology, 1998, 43, 15-24.	2.8	57
129	Expression of integrins in experimental autoimmune neuritis and guillain-barrÃ© syndrome. Annals of Neurology, 1998, 44, 611-621.	2.8	34
130	Modulation of the expression of integrins on glial cells during experimental autoimmune encephalomyelitis. A central role for TNF-alpha. American Journal of Pathology, 1997, 151, 1425-35.	1.9	29
131	Î±6Î²4 and Î±6Î²1 Integrins in Astrocytomas and Other CNS Tumors. Journal of Neuropathology and Experimental Neurology, 1996, 55, 456-465.	0.9	39
132	Î²4 integrin and other Schwann cell markers in axonal neuropathy. , 1996, 17, 294-306.		82
133	Intravenous immunoglobulin treatment in patients with chronic inflammatory demyelinating neuropathy not responsive to other treatments.. Journal of Neurology, Neurosurgery and Psychiatry, 1994, 57, 43-45.	0.9	17