Steven A Moore

List of Publications by Year in descending order

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170 papers	13,012 citations	31949 53 h-index	25770 108 g-index
175 all docs	175 docs citations	175 times ranked	15363 citing authors

#	Article	IF	Citations
1	Severe Acute Respiratory Syndrome Coronavirus Infection Causes Neuronal Death in the Absence of Encephalitis in Mice Transgenic for Human ACE2. Journal of Virology, 2008, 82, 7264-7275.	1.5	1,101
2	Post-translational disruption of dystroglycanâ€“ligand interactions in congenital muscular dystrophies. Nature, 2002, 418, 417-421.	13.7	747
3	Deletion of brain dystroglycan recapitulates aspects of congenital muscular dystrophy. Nature, 2002, 418, 422-425.	13.7	532
4	Ataluren treatment of patients with nonsense mutation dystrophinopathy. Muscle and Nerve, 2014, 50, 477-487.	1.0	357
5	Disruption of the Sarcoglycanâ€“Sarcospan Complex in Vascular Smooth Muscle. Cell, 1999, 98, 465-474.	13.5	352
6	CaMKII determines mitochondrial stress responses in heart. Nature, 2012, 491, 269-273.	13.7	340
7	Progressive Muscular Dystrophy in Î±-Sarcoglycanâ€“deficient Mice. Journal of Cell Biology, 1998, 142, 1461-1471.	2.3	331
8	Astrocytes, Not Neurons, Produce Docosahexaenoic Acid (22:6?-3) and Arachidonic Acid (20:4?-6). Journal of Neurochemistry, 1991, 56, 518-524.	2.1	308
9	Hydroxyeicosatetraenoic acids (HETEs). Progress in Lipid Research, 1988, 27, 271-323.	5.3	302
10	Diagnostic approach to the congenital muscular dystrophies. Neuromuscular Disorders, 2014, 24, 289-311.	0.3	275
11	Disruption of Dag1 in Differentiated Skeletal Muscle Reveals a Role for Dystroglycan in Muscle Regeneration. Cell, 2002, 110, 639-648.	13.5	260
12	LARGE can functionally bypass Î±-dystroglycan glycosylation defects in distinct congenital muscular dystrophies. Nature Medicine, 2004, 10, 696-703.	15.2	253
13	Rapid development of colitis in NSAID-treated IL-10â€“deficient mice. Gastroenterology, 2002, 123, 1527-1542.	0.6	252
14	Sarcolemma-localized nNOS is required to maintain activity after mild exercise. Nature, 2008, 456, 511-515.	13.7	251
15	Identification of a Functional Peroxisome Proliferator-Activated Receptor Response Element in the Rat Catalase Promoter. Molecular Endocrinology, 2002, 16, 2793-2801.	3.7	240
16	Unique Role of Dystroglycan in Peripheral Nerve Myelination, Nodal Structure, and Sodium Channel Stabilization. Neuron, 2003, 38, 747-758.	3.8	230
17	ISPD loss-of-function mutations disrupt dystroglycan O-mannosylation and cause Walker-Warburg syndrome. Nature Genetics, 2012, 44, 575-580.	9.4	212
18	Unilateral carrageenan injection into muscle or joint induces chronic bilateral hyperalgesia in rats. Pain, 2003, 104, 567-577.	2.0	207

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19	Mutations in GDP-Mannose Pyrophosphorylase B Cause Congenital and Limb-Girdle Muscular Dystrophies Associated with Hypoglycosylation of Î±-Dystroglycan. American Journal of Human Genetics, 2013, 93, 29-41.	2.6	197
20	Disruption of the Î²-Sarcoglycan Gene Reveals Pathogenetic Complexity of Limb-Girdle Muscular Dystrophy Type 2E. Molecular Cell, 2000, 5, 141-151.	4.5	185
21	Ganglioglioma. Neurosurgery, 1992, 31, 171-178.	0.6	180
22	Expanding the Boundaries of RNA Sequencing as a Diagnostic Tool for Rare Mendelian Disease. American Journal of Human Genetics, 2019, 104, 466-483.	2.6	176
23	Recessive TTN truncating mutations define novel forms of core myopathy with heart disease. Human Molecular Genetics, 2014, 23, 980-991.	1.4	149
24	Mutant lamins cause nuclear envelope rupture and DNA damage in skeletal muscle cells. Nature Materials, 2020, 19, 464-473.	13.3	148
25	Limb-Girdle Muscular Dystrophy in the United States. Journal of Neuropathology and Experimental Neurology, 2006, 65, 995-1003.	0.9	144
26	Polyunsaturated Fatty Acid Synthesis and Release. Journal of Molecular Neuroscience, 2001, 16, 195-200.	1.1	138
27	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. Nature Genetics, 2017, 49, 238-248.	9.4	131
28	Basal lamina strengthens cell membrane integrity via the laminin G domain-binding motif of Î±-dystroglycan. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 12573-12579.	3.3	125
29	Intracellular accumulation and reduced sarcolemmal expression of dysferlin in limb-girdle muscular dystrophies. Annals of Neurology, 2000, 48, 902-912.	2.8	119
30	Distinct Functions of Glial and Neuronal Dystroglycan in the Developing and Adult Mouse Brain. Journal of Neuroscience, 2010, 30, 14560-14572.	1.7	119
31	LARGE glycans on dystroglycan function as a tunable matrix scaffold to prevent dystrophy. Nature, 2013, 503, 136-140.	13.7	112
32	Role of the Blood-Brain Barrier in the Formation of Long-Chain ?-3 and ?-6 Fatty Acids from Essential Fatty Acid Precursors. Journal of Neurochemistry, 1990, 55, 391-402.	2.1	106
33	Both Laminin and Schwann Cell Dystroglycan Are Necessary for Proper Clustering of Sodium Channels at Nodes of Ranvier. Journal of Neuroscience, 2005, 25, 9418-9427.	1.7	101
34	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. Genome Biology, 2014, 15, R53.	13.9	101
35	Prevention of cardiomyopathy in mouse models lacking the smooth muscle sarcoglycan-sarcospan complex. Journal of Clinical Investigation, 2001, 107, R1-R7.	3.9	98
36	The functional O-mannose glycan on Î±-dystroglycan contains a phospho-ribitol primed for matriglycan addition. ELife, 2016, 5, .	2.8	98

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37	IL-10 Is a Central Regulator of Cyclooxygenase-2 Expression and Prostaglandin Production. Journal of Immunology, 2001, 166, 2674-2680.	0.4	85
38	ISPD gene mutations are a common cause of congenital and limb-girdle muscular dystrophies. Brain, 2013, 136, 269-281.	3.7	80
39	Dystroglycan on Radial Glia End Feet Is Required for Pial Basement Membrane Integrity and Columnar Organization of the Developing Cerebral Cortex. Journal of Neuropathology and Experimental Neurology, 2012, 71, 1047-1063.	0.9	78
40	Brain and Eye Malformations Resembling Walkerâ€“Warburg Syndrome Are Recapitulated in Mice by Dystroglycan Deletion in the Epiblast. Journal of Neuroscience, 2008, 28, 10567-10575.	1.7	77
41	Genetic ablation of complement C3 attenuates muscle pathology in dysferlin-deficient mice. Journal of Clinical Investigation, 2010, 120, 4366-4374.	3.9	77
42	A quantitative comparison of motor and sensory conduction velocities in short- and long-term streptozotocin- and alloxan-diabetic rats. Journal of the Neurological Sciences, 1980, 48, 133-152.	0.3	71
43	Congenital disorder of glycosylation due to DPM1 mutations presenting with dystroglycanopathy-type congenital muscular dystrophy. Molecular Genetics and Metabolism, 2013, 110, 345-351.	0.5	71
44	Myopathic Lamin Mutations Cause Reductive Stress and Activate the Nrf2/Keap-1 Pathway. PLoS Genetics, 2015, 11, e1005231.	1.5	71
45	Dysferlinâ€deficient muscular dystrophy features amyloidosis. Annals of Neurology, 2008, 63, 323-328.	2.8	69
46	FSH dystrophy 4q35 deletion in patients presenting with facial-sparing scapular myopathy. Neurology, 2000, 54, 1927-1931.	1.5	67
47	Smooth Muscle Can Comprise the Sarcomatous Component of Gliosarcomas. Journal of Neuropathology and Experimental Neurology, 1992, 51, 493-498.	0.9	65
48	Unusual clinical presentations in patients harboring the facioscapulohumeral dystrophy 4q35 deletion. Muscle and Nerve, 2001, 24, 352-356.	1.0	63
49	Vascular Smooth Muscle Hyperplasia Underlies the Formation of Glomeruloid Vascular Structures of Glioblastoma Multiforme. Journal of Neuropathology and Experimental Neurology, 1992, 51, 488-492.	0.9	61
50	Collagen VI glycine mutations: Perturbed assembly and a spectrum of clinical severity. Annals of Neurology, 2008, 64, 294-303.	2.8	61
51	Mutation screen reveals novel variants and expands the phenotypes associated with DYNC1H1. Journal of Neurology, 2015, 262, 2124-2134.	1.8	59
52	Skeletal Muscle Pathology in X-Linked Myotubular Myopathy: Review With Cross-Species Comparisons. Journal of Neuropathology and Experimental Neurology, 2016, 75, 102-110.	0.9	59
53	Expression of monocyte chemoattractant protein (MCP-1) and nitric oxide synthase-2 following cerebral trauma. Acta Neuropathologica, 1998, 95, 98-103.	3.9	58
54	Visual Impairment in the Absence of Dystroglycan. Journal of Neuroscience, 2009, 29, 13136-13146.	1.7	56

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55	Differential activation of catalase expression and activity by PPAR agonists: Implications for astrocyte protection in anti-glioma therapy. Redox Biology, 2013, 1, 70-79.	3.9	54
56	Novel deletion of lysine 7 expands the clinical, histopathological and genetic spectrum of TPM2-related myopathies. Brain, 2013, 136, 508-521.	3.7	53
57	Cerebral Endothelium and Astrocytes Cooperate in Supplying Docosahexaenoic Acid to Neurons. Advances in Experimental Medicine and Biology, 1993, 331, 229-233.	0.8	50
58	Allele-specific RNAi Mitigates Phenotypic Progression in a Transgenic Model of Alzheimer's Disease. Molecular Therapy, 2009, 17, 1563-1573.	3.7	48
59	Duchenne and Becker Muscular Dystrophies: A Review of Animal Models, Clinical End Points, and Biomarker Quantification. Toxicologic Pathology, 2017, 45, 961-976.	0.9	47
60	TRAPPC11 and GOSR2 mutations associate with hypoglycosylation of Î±-dystroglycan and muscular dystrophy. Skeletal Muscle, 2018, 8, 17.	1.9	47
61	Isolated Central Nervous System Angiitis First Presenting as Spontaneous Intracranial Hemorrhage. Neurosurgery, 1987, 20, 310-315.	0.6	46
62	Progressive External Ophthalmoplegia and Vision and Hearing Loss in a Patient With Mutations in POLG2 and OPA1. Archives of Neurology, 2008, 65, 125-31.	4.9	46
63	Cervical Epidural Hematoma Secondary to an Extradural Vascular Malformation in an Infant. Neurosurgery, 1995, 36, 585-588.	0.6	46
64	Antigen presentation by brain microvessel smooth muscle and endothelium. Journal of Neuroimmunology, 1990, 28, 63-71.	1.1	45
65	Expression and Vascular Effects of Cyclooxygenase-2 in Brain. Stroke, 1998, 29, 2600-2606.	1.0	45
66	Differential Opening of the Brain Endothelial Following Neutralization of the Endothelial Luminal Anionic Charge In Vitro. Journal of Neuropathology and Experimental Neurology, 1987, 46, 141-153.	0.9	44
67	Murine cerebral microvascular endothelium incorporate and metabolize 12-hydroxyeicosatetraenoic acid. Journal of Cellular Physiology, 1988, 137, 75-85.	2.0	44
68	Expression of Î³-Sarcoglycan in Smooth Muscle and Its Interaction with the Smooth Muscle Sarcoglycan-Sarcospan Complex. Journal of Biological Chemistry, 2000, 275, 38554-38560.	1.6	44
69	LMNA variants cause cytoplasmic distribution of nuclear pore proteins in Drosophila and human muscle. Human Molecular Genetics, 2012, 21, 1544-1556.	1.4	44
70	Ï‰-Oxidation of 20-Hydroxyeicosatetraenoic Acid (20-HETE) in Cerebral Microvascular Smooth Muscle and Endothelium by Alcohol Dehydrogenase 4. Journal of Biological Chemistry, 2005, 280, 33157-33164.	1.6	43
71	Muscle Fatigue Increases the Probability of Developing Hyperalgesia in Mice. Journal of Pain, 2007, 8, 692-699.	0.7	41
72	Carotid-Cavernous Sinus Thrombosis Caused by Aspergillus Fumigatus: Magnetic Resonance Imaging with Pathologic Correlationâ€"A Case Report. Angiology, 1990, 41, 652-657.	0.8	40

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73	Multiminicore Myopathy, Central Core Disease, Malignant Hyperthermia Susceptibility, and RYR1 Mutations. Archives of Neurology, 2004, 61, 27.	4.9	40
74	Astrocytes in the Rat Nucleus Tractus Solitarii Are Critical for Cardiovascular Reflex Control. Journal of Neuroscience, 2013, 33, 18608-18617.	1.7	40
75	Absence of Dystrophin Related Protein-2 disrupts Cajal bands in a patient with Charcotâ€“Marieâ€“Tooth disease. Neuromuscular Disorders, 2015, 25, 786-793.	0.3	40
76	Induction of cyclooxygenase-2 by anandamide in cerebral microvascular endothelium. Microvascular Research, 2005, 69, 28-35.	1.1	39
77	GMPPB-Associated Dystroglycanopathy: Emerging Common Variants with Phenotype Correlation. Human Mutation, 2015, 36, 1159-1163.	1.1	39
78	Reduced sensory and motor conduction velocity in 25-week-old diabetic [] mice. Experimental Neurology, 1980, 70, 548-555.	2.0	38
79	Glycogen accumulation in tibial nerves of experimentally diabetic and aging control rats. Journal of the Neurological Sciences, 1981, 52, 289-303.	0.3	38
80	Interobserver variability associated with the MIB-1 labeling index. Cancer, 2001, 92, 2720-2726.	2.0	38
81	20-Hydroxyeicosatetraenoic acid is a potent dilator of mouse basilar artery: role of cyclooxygenase. American Journal of Physiology - Heart and Circulatory Physiology, 2006, 291, H2301-H2307.	1.5	38
82	CpG DNA induces cyclooxygenase-2 expression and prostaglandin production. International Immunology, 2001, 13, 1013-1020.	1.8	37
83	Cardiac pathology exceeds skeletal muscle pathology in two cases of limbâ€girdle muscular dystrophy type 2I. Muscle and Nerve, 2009, 40, 883-889.	1.0	37
84	Receptor-Linked Hydrolysis of Phosphoinositides and Production of Prostacyclin in Cerebral Endothelial Cells. Journal of Neurochemistry, 1992, 58, 1930-1935.	2.1	36
85	Regulation of Cytokine-Induced iNOS Expression by a Hairpin Oligonucleotide in Murine Cerebral Endothelial Cells. Biochemical and Biophysical Research Communications, 1997, 235, 394-397.	1.0	36
86	Deep White Matter Pathologic Features in Watershed Regions. Archives of Neurology, 2005, 62, 1154.	4.9	36
87	Making sense of missense variants in TTN-related congenital myopathies. Acta Neuropathologica, 2021, 141, 431-453.	3.9	34
88	Primary germinoma of the spinal cord: a case report with 28-year follow-up and review of the literature. Acta Neuropathologica, 1995, 90, 657-659.	3.9	33
89	14,15-Epoxyeicosatrienoic acid inhibits prostaglandin E2 production in vascular smooth muscle cells. American Journal of Physiology - Heart and Circulatory Physiology, 1998, 275, H2113-H2121.	1.5	33
90	Limb-girdle muscular dystrophy. Current Neurology and Neuroscience Reports, 2003, 3, 78-85.	2.0	33

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91	X-linked myopathy with excessive autophagy: a failure of self-eating. Acta Neuropathologica, 2015, 129, 383-390.	3.9	33
92	Brain Microvessels Produce 12-Hydroxyeicosatetraenoic Acid. Journal of Neurochemistry, 1989, 53, 376-382.	2.1	32
93	Autosomal dominant calpainopathy due to heterozygous CAPN3 C.643_663del21. Muscle and Nerve, 2018, 57, 679-683.	1.0	32
94	Validation of Optical Genome Mapping for the Molecular Diagnosis of Facioscapulohumeral Muscular Dystrophy. Journal of Molecular Diagnostics, 2021, 23, 1506-1514.	1.2	32
95	Cerebral microvascular smooth muscle in tissue culture. In Vitro, 1984, 20, 512-520.	1.2	31
96	Myelinoclastic Diffuse Sclerosis (Schilder's Disease): Report of a Case and Review of the Literature. Journal of Child Neurology, 1994, 9, 398-403.	0.7	31
97	Further evidence of Fukutin mutations as a cause of childhood onset limb-girdle muscular dystrophy without mental retardation. Neuromuscular Disorders, 2009, 19, 352-356.	0.3	31
98	Glial scaffold required for cerebellar granule cell migration is dependent on dystroglycan function as a receptor for basement membrane proteins. Acta Neuropathologica Communications, 2013, 1, 58.	2.4	31
99	Regional localization of virus in the central nervous system of mice persistently infected with murine coronavirus JHM. Virology, 1988, 166, 328-338.	1.1	30
100	A common disease-associated missense mutation in alpha-sarcoglycan fails to cause muscular dystrophy in mice. Human Molecular Genetics, 2008, 17, 1201-1213.	1.4	30
101	Infantile onset CMT2D/dSMA V in monozygotic twins due to a mutation in the anticodonâ€binding domain of GARS. Journal of the Peripheral Nervous System, 2012, 17, 132-134.	1.4	28
102	SMCHD1 mutation spectrum for facioscapulohumeral muscular dystrophy type 2 (FSHD2) and Bosma arhinia microphthalmia syndrome (BAMS) reveals disease-specific localisation of variants in the ATPase domain. Journal of Medical Genetics, 2019, 56, 693-700.	1.5	27
103	What Every Neuropathologist Needs to Know: The Muscle Biopsy. Journal of Neuropathology and Experimental Neurology, 2020, 79, 719-733.	0.9	25
104	Founder Fukutin mutation causes Walkerâ€“Warburg syndrome in four Ashkenazi Jewish families. Prenatal Diagnosis, 2009, 29, 560-569.	1.1	24
105	FSHD type 2 and Bosma arhinia microphthalmia syndrome. Neurology, 2018, 91, e562-e570.	1.5	24
106	Cellular and Vessel Wall Morphology of Cerebral Cortical Arterioles after Short-Term Diabetes in Adult Rats. Journal of Vascular Research, 1985, 22, 265-277.	0.6	23
107	Presentation, Management and Follow-Up of Schilderâ€™s Disease. Pediatric Neurosurgery, 1998, 29, 86-91.	0.4	23
108	Cardiac damage after lesions of the nucleus tractus solitarii. American Journal of Physiology - Regulatory Integrative and Comparative Physiology, 2009, 296, R272-R279.	0.9	22

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109	Elevated Oxidative Membrane Damage Associated with Genetic Modifiers of Lyst-Mutant Phenotypes. PLoS Genetics, 2010, 6, e1001008.	1.5	22
110	Brain Micro vessel 12-Hydroxyeicosatetraenoic Acid Is the (S) Enantiomer and Is Lipoxygenase Derived. Journal of Neurochemistry, 1991, 57, 922-929.	2.1	21
111	Reduced neurofilament expression in cutaneous nerve fibers of patients with CMT2E. Neurology, 2015, 85, 228-234.	1.5	21
112	Uniparental disomy unveils a novel recessive mutation in POMT2. Neuromuscular Disorders, 2018, 28, 592-596.	0.3	20
113	Ultrastructural axonal pathology in experimentally diabetic and aging control rats. Brain Research Bulletin, 1982, 8, 317-323.	1.4	19
114	Endogenous Glucuronyltransferase Activity of LARGE or LARGE2 Required for Functional Modification of Î±-Dystroglycan in Cells and Tissues. Journal of Biological Chemistry, 2014, 289, 28138-28148.	1.6	19
115	Acute and chronic exposure of mouse cerebral microvessel endothelial cells to increased concentrations of glucose and galactose: Effect on myo-inositol metabolism, PGE2 synthesis, and transport activity. Metabolism: Clinical and Experimental, 1991, 40, 347-358.	1.5	18
116	Evaluation of commercial dysferlin antibodies on canine, mouse and human skeletal muscle. Neuromuscular Disorders, 2010, 20, 820-825.	0.3	18
117	CLIA Laboratory Testing for Facioscapulohumeral Dystrophy. Neurology, 2021, 96, e1054-e1062.	1.5	18
118	Late adultâ€onset of Xâ€linked myopathy with excessive autophagy. Muscle and Nerve, 2014, 50, 138-144.	1.0	17
119	Early-progressive dilated cardiomyopathy in a family with Becker muscular dystrophy related to a novel frameshift mutation in the dystrophin gene exon 27. Journal of Human Genetics, 2015, 60, 151-155.	1.1	17
120	Inclusion body myositis and sarcoid myopathy: Coincidental occurrence or associated diseases. Neuromuscular Disorders, 2015, 25, 297-300.	0.3	17
121	Studies of hyperlipidemia in drug-induced diabetic rats by high-performance liquid chromatography. Biomedical Applications, 1980, 221, 19-26.	1.7	16
122	Rare Manifestation of a c.290 C>T, p.Gly97GluVCPMutation. Case Reports in Genetics, 2015, 2015, 1-5.	0.1	16
123	Intrafamilial variability in GMPPB -associated dystroglycanopathy: Broadening of the phenotype. Neurology, 2015, 84, 1495-1497.	1.5	16
124	Suppression of myopathic lamin mutations by muscle-specific activation ofAMPKand modulation of downstream signaling. Human Molecular Genetics, 2019, 28, 351-371.	1.4	16
125	Intron mutations and early transcription termination in Duchenne and Becker muscular dystrophy. Human Mutation, 2022, 43, 511-528.	1.1	16
126	Uptake of Adenosine by Cultured Cerebral Vascular Smooth Muscle Cells. Journal of Neurochemistry, 1983, 41, 939-941.	2.1	15

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127	Novel Mutation in Spectrin-like Repeat 1 of Dystrophin Central Domain Causes Protein Misfolding and Mild Becker Muscular Dystrophy. Journal of Biological Chemistry, 2012, 287, 18153-18162.	1.6	15
128	Cytoplasmic body pathology in severe ACTA1 -related myopathy in the absence of typical nemaline rods. Neuromuscular Disorders, 2017, 27, 531-536.	0.3	15
129	Regulation of prostaglandin H synthase-2 expression in cerebromicrovascular smooth muscle by serum and epidermal growth factor. , 1998, 176, 495-505.		14
130	A novel missense mutation in POMT1 modulates the severe congenital muscular dystrophy phenotype associated with POMT1 nonsense mutations. Neuromuscular Disorders, 2014, 24, 312-320.	0.3	14
131	Variable disease severity in Saudi Arabian and Sudanese families with c.3924 + 2 T > C mutation of LAMA2. BMC Research Notes, 2011, 4, 534.	0.6	13
132	Aberrant regulation of epigenetic modifiers contributes to the pathogenesis in patients with selenoprotein N â€ related myopathies. Human Mutation, 2019, 40, 962-974.	1.1	13
133	Chymopapain-induced reduction of proinflammatory phospholipase A2 activity and amelioration of neuropathic behavioral changes in an in vivo model of acute sciatica. Journal of Neurosurgery, 1997, 86, 998-1006.	0.9	12
134	A novel ANO5 splicing variant in a LGMD2L patient leads to production of a truncated aggregationâ€prone Ano5 peptide. Journal of Pathology: Clinical Research, 2018, 4, 135-145.	1.3	12
135	The inflammatory pathology of dysferlinopathy is distinct from calpainopathy, Becker muscular dystrophy, and inflammatory myopathies. Acta Neuropathologica Communications, 2022, 10, 17.	2.4	12
136	Sudden Death and Myocardial Lesions after Damage to Catecholamine Neurons of the Nucleus Tractus Solitarii in Rat. Cellular and Molecular Neurobiology, 2012, 32, 1119-1126.	1.7	11
137	A novel CAPN3 mutation in late-onset limb-girdle muscular dystrophy with early respiratory insufficiency. Journal of Clinical Neuroscience, 2018, 53, 229-231.	0.8	11
138	Absence of Axoglial Paranodal Junctions in a Child With CNTNAP1 Mutations, Hypomyelination, and Arthrogryposis. Journal of Child Neurology, 2018, 33, 642-650.	0.7	11
139	Clinical, genetic, and pathologic characterization of FKRP Mexican founder mutation c.1387A>G. Neurology: Genetics, 2019, 5, e315.	0.9	11
140	Validation of a Muscle-Specific Tissue Image Analysis Tool for Quantitative Assessment of Dystrophin Staining in Frozen Muscle Biopsies. Archives of Pathology and Laboratory Medicine, 2019, 143, 197-205.	1.2	11
141	Pathogenic variants in TNNC2 cause congenital myopathy due to an impaired force response to calcium. Journal of Clinical Investigation, 2021, 131, .	3.9	11
142	Optic Neuritis: A Novel Presentation of Schilder's Disease. Journal of Child Neurology, 2001, 16, 693-696.	0.7	10
143	A slowly progressive form of limbâ€girdle muscular dystrophy type 2C associated with founder mutation in the SGCG gene in Puerto Rican Hispanics. Molecular Genetics & Genomic Medicine, 2015, 3, 92-98.	0.6	10
144	Arginine Vasopressin Stimulates Protein Synthesis but Not Proliferation of Cultured Vascular Endothelial Cells. Journal of Cardiovascular Pharmacology, 1995, 25, 368-375.	0.8	9

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145	Joiner et al. reply. Nature, 2014, 513, E3-E3.	13.7	9
146	The Oxygen Radical Scavenger Pyrrolidine Dithiocarbamate Enhances Interleukin-1Î²-Induced Cyclooxygenase-2 Expression in Cerebral Microvascular Smooth Muscle Cells. Microvascular Research, 2002, 64, 405-413.	1.1	8
147	Metabolism of anandamide in cerebral microvascular endothelial cells. Prostaglandins and Other Lipid Mediators, 2004, 73, 59-72.	1.0	8
148	Congenital myasthenic syndrome caused by a frameshift insertion mutation in GFPT1. Neurology: Genetics, 2020, 6, e468.	0.9	8
149	Primary germinoma of the spinal cord: a case report with 28-year follow-up and review of the literature. Acta Neuropathologica, 1995, 90, 657-659.	3.9	8
150	Urologic and gastrointestinal symptoms in the dystroglycanopathies. Neurology, 2015, 84, 532-539.	1.5	7
151	Schwann cell heterogeneityâ€"Might it underlie the diversity seen in schwann cell tumors?. Human Pathology, 1994, 25, 1113.	1.1	6
152	Dystrophinopathy presenting with arrhythmia in an asymptomatic 34-year-old man: a case report. Journal of Medical Case Reports, 2009, 3, 8625.	0.4	6
153	Magnetic Resonance Imaging characteristics in case of TOR1AIP1 muscular dystrophy. Clinical Imaging, 2019, 58, 108-113.	0.8	6
154	Cobblestone Malformation in LAMA2 Congenital Muscular Dystrophy (MDC1A). Journal of Neuropathology and Experimental Neurology, 2020, 79, 998-1010.	0.9	6
155	Limb-girdle muscular dystrophies. Advances in Neurology, 2002, 88, 273-91.	0.8	6
156	ACTA1-myopathy with prominent finger flexor weakness and rimmed vacuoles. Neuromuscular Disorders, 2019, 29, 388-391.	0.3	5
157	Hughlings Jackson and the role of the entorhinal cortex in temporal lobe epilepsy: From Patient A to Doctor Z. Epilepsy and Behavior, 2006, 9, 524-531.	0.9	4
158	Differential Metabolism of Hydroxyeicosatetraenoic Acid Isomers by Mouse Cerebromicrovascular Endothelium. Journal of Neurochemistry, 1992, 58, 374-382.	2.1	3
159	Child Neurology: LAMA2 muscular dystrophy without contractures. Neurology, 2017, 88, e199-e203.	1.5	3
160	Dystrophinopathy muscle biopsies in the genetic testing ERA: One center's data. Muscle and Nerve, 2018, 58, 149-153.	1.0	3
161	Cross-sectional Neuromuscular Phenotyping Study of Patients With Arhinia With SMCHD1 Variants. Neurology, 2022, 98, .	1.5	3
162	Thalamic ependymoma presenting as recurrent subarachnoid hemorrhage. Journal of Stroke and Cerebrovascular Diseases, 1992, 2, 106-109.	0.7	2

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163	Clinical Reasoning: A 30-year-old man with progressive weakness and atrophy. Neurology, 2016, 87, e227-e230.	1.5	2
164	A novel noncoding FKRP mutation in early onset limb-girdle muscular dystrophy. Neurology: Genetics, 2020, 6, e388.	0.9	1
165	Refractory infantile highâ€grade glioma containing TRKâ€fusion responds to larotrectinib. Pediatric Blood and Cancer, 2021, 68, e28868.	0.8	1
166	Sphingosine Phosphate Lyase Is Upregulated in Duchenne Muscular Dystrophy, and Its Inhibition Early in Life Attenuates Inflammation and Dystrophy in Mdx Mice. International Journal of Molecular Sciences, 2022, 23, 7579.	1.8	1
167	Eicosanoid Production by Isolated Cerebral Microvessels and Cultured Cerebral Endothelium. Annals of the New York Academy of Sciences, 1989, 559, 471-473.	1.8	0
168	Congenital Myasthenic Syndromes. , 2021, , 313-316.		0
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