Steven A Moore

List of Publications by Citations

167 papers	10,645 citations	49 h-index	100 g-index
175 ext. papers	11,896 ext. citations	8.3 avg, IF	5.6 L-index

#	Paper	IF	Citations
167	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, 7264-75	6.6	829
166	Post-translational disruption of dystroglycan-ligand interactions in congenital muscular dystrophies. *Nature*, **2002**, 418, 417-22	50.4	667
165	Deletion of brain dystroglycan recapitulates aspects of congenital muscular dystrophy. *Nature*, **2002**, 418, 422-5	50.4	473
164	Disruption of the sarcoglycan-sarcospan complex in vascular smooth muscle: a novel mechanism for cardiomyopathy and muscular dystrophy. *Cell*, **1999**, 98, 465-74	56.2	321
163	Progressive muscular dystrophy in alpha-sarcoglycan-deficient mice. *Journal of Cell Biology*, **1998**, 142, 1461-71	7.3	303
162	CaMKII determines mitochondrial stress responses in heart. *Nature*, **2012**, 491, 269-73	50.4	290
161	Ataluren treatment of patients with nonsense mutation dystrophinopathy. *Muscle and Nerve*, **2014**, 50, 477-87	3.4	281
160	Astrocytes, not neurons, produce docosahexaenoic acid (22:6 omega-3) and arachidonic acid (20:4 omega-6). *Journal of Neurochemistry*, **1991**, 56, 518-24	6	276
159	Hydroxyeicosatetraenoic acids (HETEs). *Progress in Lipid Research*, **1988**, 27, 271-323	14.3	270
158	Rapid development of colitis in NSAID-treated IL-10-deficient mice. *Gastroenterology*, **2002**, 123, 1527-42	13.3	233
157	Sarcolemma-localized nNOS is required to maintain activity after mild exercise. *Nature*, **2008**, 456, 511-5	50.4	232
156	Disruption of DAG1 in differentiated skeletal muscle reveals a role for dystroglycan in muscle regeneration. *Cell*, **2002**, 110, 639-48	56.2	218
155	Diagnostic approach to the congenital muscular dystrophies. *Neuromuscular Disorders*, **2014**, 24, 289-311	2.9	217
154	Identification of a functional peroxisome proliferator-activated receptor response element in the rat catalase promoter. *Molecular Endocrinology*, **2002**, 16, 2793-801		217
153	LARGE can functionally bypass alpha-dystroglycan glycosylation defects in distinct congenital muscular dystrophies. *Nature Medicine*, **2004**, 10, 696-703	50.5	215
152	Unique role of dystroglycan in peripheral nerve myelination, nodal structure, and sodium channel stabilization. *Neuron*, **2003**, 38, 747-58	13.9	208
151	ISPD loss-of-function mutations disrupt dystroglycan O-mannosylation and cause Walker-Warburg syndrome. *Nature Genetics*, **2012**, 44, 575-80	36.3	183

150	Unilateral carrageenan injection into muscle or joint induces chronic bilateral hyperalgesia in rats. *Pain*, **2003**, 104, 567-577	8	178
149	Disruption of the beta-sarcoglycan gene reveals pathogenetic complexity of limb-girdle muscular dystrophy type 2E. *Molecular Cell*, **2000**, 5, 141-51	17.6	167
148	Mutations in GDP-mannose pyrophosphorylase B cause congenital and limb-girdle muscular dystrophies associated with hypoglycosylation of □dystroglycan. *American Journal of Human Genetics*, **2013**, 93, 29-41	11	162
147	Ganglioglioma: 13 years of experience. *Neurosurgery*, **1992**, 31, 171-8	3.2	161
146	Limb-girdle muscular dystrophy in the United States. *Journal of Neuropathology and Experimental Neurology*, **2006**, 65, 995-1003	3.1	118
145	Recessive TTN truncating mutations define novel forms of core myopathy with heart disease. *Human Molecular Genetics*, **2014**, 23, 980-91	5.6	117
144	Polyunsaturated fatty acid synthesis and release by brain-derived cells in vitro. *Journal of Molecular Neuroscience*, **2001**, 16, 195-200; discussion 215-21	3.3	111
143	Expanding the Boundaries of RNA Sequencing as a Diagnostic Tool for Rare Mendelian Disease. *American Journal of Human Genetics*, **2019**, 104, 466-483	11	110
142	Intracellular accumulation and reduced sarcolemmal expression of dysferlin in limb□girdle muscular dystrophies. *Annals of Neurology*, **2000**, 48, 902-912	9.4	107
141	Basal lamina strengthens cell membrane integrity via the laminin G domain-binding motif of alpha-dystroglycan. *Proceedings of the National Academy of Sciences of the United States of America*, **2009**, 106, 12573-9	11.5	106
140	Role of the blood-brain barrier in the formation of long-chain omega-3 and omega-6 fatty acids from essential fatty acid precursors. *Journal of Neurochemistry*, **1990**, 55, 391-402	6	97
139	Both laminin and Schwann cell dystroglycan are necessary for proper clustering of sodium channels at nodes of Ranvier. *Journal of Neuroscience*, **2005**, 25, 9418-27	6.6	91
138	Distinct functions of glial and neuronal dystroglycan in the developing and adult mouse brain. *Journal of Neuroscience*, **2010**, 30, 14560-72	6.6	90
137	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. *Nature Genetics*, **2017**, 49, 238-248	36.3	88
136	Prevention of cardiomyopathy in mouse models lacking the smooth muscle sarcoglycan-sarcospan complex. *Journal of Clinical Investigation*, **2001**, 107, R1-7	15.9	88
135	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	18.3	86
134	LARGE glycans on dystroglycan function as a tunable matrix scaffold to prevent dystrophy. *Nature*, **2013**, 503, 136-40	50.4	79
133	Mutant lamins cause nuclear envelope rupture and DNA damage in skeletal muscle cells. *Nature Materials*, **2020**, 19, 464-473	27	76

132	The functional O-mannose glycan on □dystroglycan contains a phospho-ribitol primed for matriglycan addition. *ELife*, **2016**, 5,	8.9	73
131	Il-10 is a central regulator of cyclooxygenase-2 expression and prostaglandin production. *Journal of Immunology*, **2001**, 166, 2674-80	5.3	72
130	Brain and eye malformations resembling Walker-Warburg syndrome are recapitulated in mice by dystroglycan deletion in the epiblast. *Journal of Neuroscience*, **2008**, 28, 10567-75	6.6	66
129	ISPD gene mutations are a common cause of congenital and limb-girdle muscular dystrophies. *Brain* , **2013**, 136, 269-81	11.2	65
128	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, 133-52	3.2	65
127	Genetic ablation of complement C3 attenuates muscle pathology in dysferlin-deficient mice. *Journal of Clinical Investigation*, **2010**, 120, 4366-74	15.9	65
126	Congenital disorder of glycosylation due to DPM1 mutations presenting with dystroglycanopathy-type congenital muscular dystrophy. *Molecular Genetics and Metabolism*, **2013**, 110, 345-351	3.7	60
125	Myopathic lamin mutations cause reductive stress and activate the nrf2/keap-1 pathway. *PLoS Genetics*, **2015**, 11, e1005231	6	59
124	FSH dystrophy 4q35 deletion in patients presenting with facial-sparing scapular myopathy. *Neurology*, **2000**, 54, 1927-31	6.5	59
123	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, 1047-63	3.1	56
122	Vascular smooth muscle hyperplasia underlies the formation of glomeruloid vascular structures of glioblastoma multiforme. *Journal of Neuropathology and Experimental Neurology*, **1992**, 51, 488-92	3.1	55
121	Unusual clinical presentations in patients harboring the facioscapulohumeral dystrophy 4q35 deletion. *Muscle and Nerve*, **2001**, 24, 352-6	3.4	52
120	Expression of monocyte chemoattractant protein (MCP-1) and nitric oxide synthase-2 following cerebral trauma. *Acta Neuropathologica*, **1998**, 95, 98-103	14.3	51
119	Dysferlin-deficient muscular dystrophy features amyloidosis. *Annals of Neurology*, **2008**, 63, 323-8	9.4	51
118	Collagen VI glycine mutations: perturbed assembly and a spectrum of clinical severity. *Annals of Neurology*, **2008**, 64, 294-303	9.4	49
117	Smooth muscle can comprise the sarcomatous component of gliosarcomas. *Journal of Neuropathology and Experimental Neurology*, **1992**, 51, 493-8	3.1	49
116	Mutation screen reveals novel variants and expands the phenotypes associated with DYNC1H1. *Journal of Neurology*, **2015**, 262, 2124-34	5.5	46
115	Differential activation of catalase expression and activity by PPAR agonists: implications for astrocyte protection in anti-glioma therapy. *Redox Biology*, **2013**, 1, 70-9	11.3	45

114	Visual impairment in the absence of dystroglycan. *Journal of Neuroscience*, **2009**, 29, 13136-46	6.6	45
113	Novel deletion of lysine 7 expands the clinical, histopathological and genetic spectrum of TPM2-related myopathies. *Brain*, **2013**, 136, 508-21	11.2	43
112	Cerebral endothelium and astrocytes cooperate in supplying docosahexaenoic acid to neurons. *Advances in Experimental Medicine and Biology*, **1993**, 331, 229-33	3.6	43
111	Progressive external ophthalmoplegia and vision and hearing loss in a patient with mutations in POLG2 and OPA1. *Archives of Neurology*, **2008**, 65, 125-31		42
110	Antigen presentation by brain microvessel smooth muscle and endothelium. *Journal of Neuroimmunology*, **1990**, 28, 63-71	3.5	42
109	Isolated central nervous system angiitis first presenting as spontaneous intracranial hemorrhage. *Neurosurgery*, **1987**, 20, 310-5	3.2	42
108	Allele-specific RNAi mitigates phenotypic progression in a transgenic model of Alzheimer's disease. *Molecular Therapy*, **2009**, 17, 1563-73	11.7	41
107	Expression and vascular effects of cyclooxygenase-2 in brain. *Stroke*, **1998**, 29, 2600-6	6.7	41
106	Differential opening of the brain endothelial barrier following neutralization of the endothelial luminal anionic charge in vitro. *Journal of Neuropathology and Experimental Neurology*, **1987**, 46, 141-53	3.1	40
105	Murine cerebral microvascular endothelium incorporate and metabolize 12-hydroxyeicosatetraenoic acid. *Journal of Cellular Physiology*, **1988**, 137, 75-85	7	40
104	Muscle fatigue increases the probability of developing hyperalgesia in mice. *Journal of Pain*, **2007**, 8, 692-9	5.2	39
103	Omega-oxidation of 20-hydroxyeicosatetraenoic acid (20-HETE) in cerebral microvascular smooth muscle and endothelium by alcohol dehydrogenase 4. *Journal of Biological Chemistry*, **2005**, 280, 33157-64	5.4	37
102	Expression of gamma -sarcoglycan in smooth muscle and its interaction with the smooth muscle sarcoglycan-sarcospan complex. *Journal of Biological Chemistry*, **2000**, 275, 38554-60	5.4	37
101	Carotid-cavernous sinus thrombosis caused by Aspergillus fumigatus: magnetic resonance imaging with pathologic correlation--a case report. *Angiology*, **1990**, 41, 652-7	2.1	37
100	Glycogen accumulation in tibial nerves of experimentally diabetic and aging control rats. *Journal of the Neurological Sciences*, **1981**, 52, 289-303	3.2	37
99	Regulation of cytokine-induced iNOS expression by a hairpin oligonucleotide in murine cerebral endothelial cells. *Biochemical and Biophysical Research Communications*, **1997**, 235, 394-7	3.4	36
98	20-Hydroxyeicosatetraenoic acid is a potent dilator of mouse basilar artery: role of cyclooxygenase. *American Journal of Physiology - Heart and Circulatory Physiology*, **2006**, 291, H2301-7	5.2	36
97	Induction of cyclooxygenase-2 by anandamide in cerebral microvascular endothelium. *Microvascular Research*, **2005**, 69, 28-35	3.7	36

96	CpG DNA induces cyclooxygenase-2 expression and prostaglandin production. *International Immunology*, **2001**, 13, 1013-20	4.9	36
95	Skeletal Muscle Pathology in X-Linked Myotubular Myopathy: Review With Cross-Species Comparisons. *Journal of Neuropathology and Experimental Neurology*, **2016**, 75, 102-10	3.1	35
94	Multiminicore myopathy, central core disease, malignant hyperthermia susceptibility, and RYR1 mutations: one disease with many faces?. *Archives of Neurology*, **2004**, 61, 27-9		35
93	Reduced sensory and motor conduction velocity in 25-week-old diabetic [C57BL/Ks (db/db)] mice. *Experimental Neurology*, **1980**, 70, 548-55	5.7	35
92	Cervical epidural hematoma secondary to an extradural vascular malformation in an infant: case report. *Neurosurgery*, **1995**, 36, 585-7; discussion 587-8	3.2	35
91	Interobserver variability associated with the MIB-1 labeling index: high levels suggest limited prognostic usefulness for patients with primary brain tumors. *Cancer*, **2001**, 92, 2720-6	6.4	34
90	LMNA variants cause cytoplasmic distribution of nuclear pore proteins in Drosophila and human muscle. *Human Molecular Genetics*, **2012**, 21, 1544-56	5.6	33
89	Receptor-linked hydrolysis of phosphoinositides and production of prostacyclin in cerebral endothelial cells. *Journal of Neurochemistry*, **1992**, 58, 1930-5	6	33
88	Cardiac pathology exceeds skeletal muscle pathology in two cases of limb-girdle muscular dystrophy type 2I. *Muscle and Nerve*, **2009**, 40, 883-9	3.4	32
87	Primary germinoma of the spinal cord: a case report with 28-year follow-up and review of the literature. *Acta Neuropathologica*, **1995**, 90, 657-9	14.3	32
86	Brain microvessels produce 12-hydroxyeicosatetraenoic acid. *Journal of Neurochemistry*, **1989**, 53, 376-82		32
85	GMPPB-Associated Dystroglycanopathy: Emerging Common Variants with Phenotype Correlation. *Human Mutation*, **2015**, 36, 1159-63	4.7	31
84	Deep white matter pathologic features in watershed regions: a novel pattern of central nervous system involvement in MELAS. *Archives of Neurology*, **2005**, 62, 1154-6		31
83	Cerebral microvascular smooth muscle in tissue culture. *In Vitro*, **1984**, 20, 512-20		31
82	Astrocytes in the rat nucleus tractus solitarii are critical for cardiovascular reflex control. *Journal of Neuroscience*, **2013**, 33, 18608-17	6.6	30
81	Duchenne and Becker Muscular Dystrophies: A Review of Animal Models, Clinical End Points, and Biomarker Quantification. *Toxicologic Pathology*, **2017**, 45, 961-976	2.1	29
80	TRAPPC11 and GOSR2 mutations associate with hypoglycosylation of dystroglycan and muscular dystrophy. *Skeletal Muscle*, **2018**, 8, 17	5.1	29
79	14,15-Epoxyeicosatrienoic acid inhibits prostaglandin E2 production in vascular smooth muscle cells. *American Journal of Physiology - Heart and Circulatory Physiology*, **1998**, 275, H2113-21	5.2	26

78	A common disease-associated missense mutation in alpha-sarcoglycan fails to cause muscular dystrophy in mice. *Human Molecular Genetics*, **2008**, 17, 1201-13	5.6	25
77	Regional localization of virus in the central nervous system of mice persistently infected with murine coronavirus JHM. *Virology*, **1988**, 166, 328-38	3.6	25
76	X-linked myopathy with excessive autophagy: a failure of self-eating. *Acta Neuropathologica*, **2015**, 129, 383-90	14.3	24
75	Limb-girdle muscular dystrophy. *Current Neurology and Neuroscience Reports*, **2003**, 3, 78-85	6.6	24
74	Infantile onset CMT2D/dSMA V in monozygotic twins due to a mutation in the anticodon-binding domain of GARS. *Journal of the Peripheral Nervous System*, **2012**, 17, 132-4	4.7	22
73	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	7.3	22
72	Myelinoclastic diffuse sclerosis (Schilder disease): report of a case and review of the literature. *Journal of Child Neurology*, **1994**, 9, 398-403	2.5	22
71	Elevated oxidative membrane damage associated with genetic modifiers of Lyst-mutant phenotypes. *PLoS Genetics*, **2010**, 6, e1001008	6	20
70	Cardiac damage after lesions of the nucleus tractus solitarii. *American Journal of Physiology - Regulatory Integrative and Comparative Physiology*, **2009**, 296, R272-9	3.2	20
69	Further evidence of Fukutin mutations as a cause of childhood onset limb-girdle muscular dystrophy without mental retardation. *Neuromuscular Disorders*, **2009**, 19, 352-6	2.9	20
68	Presentation, management and follow-up of Schilder disease. *Pediatric Neurosurgery*, **1998**, 29, 86-91	0.9	20
67	Brain microvessel 12-hydroxyeicosatetraenoic acid is the (S) enantiomer and is lipoxygenase derived. *Journal of Neurochemistry*, **1991**, 57, 922-9	6	20
66	Ultrastructural axonal pathology in experimentally diabetic and aging control rats. *Brain Research Bulletin*, **1982**, 8, 317-23	3.9	19
65	Autosomal dominant calpainopathy due to heterozygous CAPN3 C.643_663del21. *Muscle and Nerve* , **2018**, 57, 679-683	3.4	18
64	Absence of Dystrophin Related Protein-2 disrupts Cajal bands in a patient with Charcot-Marie-Tooth disease. *Neuromuscular Disorders*, **2015**, 25, 786-93	2.9	17
63	Founder Fukutin mutation causes Walker-Warburg syndrome in four Ashkenazi Jewish families. *Prenatal Diagnosis*, **2009**, 29, 560-9	3.2	17
62	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 Na+/K(+)-ATPase transport activity. *Metabolism: Clinical and Experimental*, **1991**, 40, 347-58	12.7	17
61	FSHD type 2 and Bosma arhinia microphthalmia syndrome: Two faces of the same mutation. *Neurology*, **2018**, 91, e562-e570	6.5	16

60	Reduced neurofilament expression in cutaneous nerve fibers of patients with CMT2E. *Neurology*, **2015**, 85, 228-34	6.5	16
59	Studies of hyperlipidemia in drug-induced diabetic rats by high-performance liquid chromatography. *Biomedical Applications*, **1980**, 221, 19-26		16
58	What Every Neuropathologist Needs to Know: The Muscle Biopsy. *Journal of Neuropathology and Experimental Neurology*, **2020**, 79, 719-733	3.1	15
57	Uptake of adenosine by cultured cerebral vascular smooth muscle cells. *Journal of Neurochemistry*, **1983**, 41, 939-41	6	15
56	Intrafamilial variability in GMPPB-associated dystroglycanopathy: Broadening of the phenotype. *Neurology*, **2015**, 84, 1495-7	6.5	14
55	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	5.8	14
54	Evaluation of commercial dysferlin antibodies on canine, mouse and human skeletal muscle. *Neuromuscular Disorders*, **2010**, 20, 820-5	2.9	14
53	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, 18153-62	5.4	14
52	Cellular and vessel wall morphology of cerebral cortical arterioles after short-term diabetes in adult rats. *Journal of Vascular Research*, **1985**, 22, 265-77	1.9	14
51	Rare Manifestation of a c.290 C>T, p.Gly97Glu VCP Mutation. *Case Reports in Genetics*, **2015**, 2015, 239167	7.7	13
50	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, 151-5	4.3	13
49	Inclusion body myositis and sarcoid myopathy: coincidental occurrence or associated diseases. *Neuromuscular Disorders*, **2015**, 25, 297-300	2.9	12
48	A novel missense mutation in POMT1 modulates the severe congenital muscular dystrophy phenotype associated with POMT1 nonsense mutations. *Neuromuscular Disorders*, **2014**, 24, 312-20	2.9	12
47	Late adult-onset of X-linked myopathy with excessive autophagy. *Muscle and Nerve*, **2014**, 50, 138-44	3.4	12
46	Endogenous glucuronyltransferase activity of LARGE or LARGE2 required for functional modification of □dystroglycan in cells and tissues. *Journal of Biological Chemistry*, **2014**, 289, 28138-48	5.4	10
45	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	3.2	10
44	Uniparental disomy unveils a novel recessive mutation in POMT2. *Neuromuscular Disorders*, **2018**, 28, 592-596	2.9	9
43	Sudden death and myocardial lesions after damage to catecholamine neurons of the nucleus tractus solitarii in rat. *Cellular and Molecular Neurobiology*, **2012**, 32, 1119-26	4.6	9

42	Regulation of prostaglandin H synthase-2 expression in cerebromicrovascular smooth muscle by serum and epidermal growth factor. *Journal of Cellular Physiology*, **1998**, 176, 495-505	7	9
41	Arginine vasopressin stimulates protein synthesis but not proliferation of cultured vascular endothelial cells. *Journal of Cardiovascular Pharmacology*, **1995**, 25, 368-75	3.1	9
40	A novel ANO5 splicing variant in a LGMD2L patient leads to production of a truncated aggregation-prone Ano5 peptide. *Journal of Pathology: Clinical Research*, **2018**, 4, 135-145	5.3	8
39	Absence of Axoglial Paranodal Junctions in a Child With CNTNAP1 Mutations, Hypomyelination, and Arthrogryposis. *Journal of Child Neurology*, **2018**, 33, 642-650	2.5	8
38	Metabolism of anandamide in cerebral microvascular endothelial cells. *Prostaglandins and Other Lipid Mediators*, **2004**, 73, 59-72	3.7	8
37	The oxygen radical scavenger pyrrolidine dithiocarbamate enhances interleukin-1beta-induced cyclooxygenase-2 expression in cerebral microvascular smooth muscle cells. *Microvascular Research* , **2002**, 64, 405-13	3.7	8
36	Cytoplasmic body pathology in severe ACTA1-related myopathy in the absence of typical nemaline rods. *Neuromuscular Disorders*, **2017**, 27, 531-536	2.9	7
35	Clinical, genetic, and pathologic characterization of Mexican founder mutation c.1387A>G. *Neurology: Genetics*, **2019**, 5, e315	3.8	7
34	A novel CAPN3 mutation in late-onset limb-girdle muscular dystrophy with early respiratory insufficiency. *Journal of Clinical Neuroscience*, **2018**, 53, 229-231	2.2	7
33	Joiner et al. reply. *Nature*, **2014**, 513, E3	50.4	7
32	Variable disease severity in Saudi Arabian and Sudanese families with c.3924 + 2 T > C mutation of LAMA2. *BMC Research Notes*, **2011**, 4, 534	2.3	7
31	Optic neuritis: a novel presentation of Schilder disease. *Journal of Child Neurology*, **2001**, 16, 693-6	2.5	7
30	Suppression of myopathic lamin mutations by muscle-specific activation of AMPK and modulation of downstream signaling. *Human Molecular Genetics*, **2019**, 28, 351-371	5.6	7
29	Aberrant regulation of epigenetic modifiers contributes to the pathogenesis in patients with selenoprotein N-related myopathies. *Human Mutation*, **2019**, 40, 962-974	4.7	6
28	Making sense of missense variants in TTN-related congenital myopathies. *Acta Neuropathologica*, **2021**, 141, 431-453	14.3	6
27	Urologic and gastrointestinal symptoms in the dystroglycanopathies. *Neurology*, **2015**, 84, 532-9	6.5	5
26	A slowly progressive form of limb-girdle muscular dystrophy type 2C associated with founder mutation in the SGCG gene in Puerto Rican Hispanics. *Molecular Genetics & Genomic Medicine*, **2015**, 3, 92-8	2.3	5
25	CLIA Laboratory Testing for Facioscapulohumeral Dystrophy: A Retrospective Analysis. *Neurology*, **2021**, 96, e1054-e1062	6.5	5

24	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	5	5
23	Limb-girdle muscular dystrophies. *Advances in Neurology*, **2002**, 88, 273-91		5
22	Magnetic Resonance Imaging characteristics in case of TOR1AIP1 muscular dystrophy. *Clinical Imaging*, **2019**, 58, 108-113	2.7	4
21	Dystrophinopathy presenting with arrhythmia in an asymptomatic 34-year-old man: a case report. *Journal of Medical Case Reports*, **2009**, 3, 8625	1.2	4
20	Hughlings Jackson and the role of the entorhinal cortex in temporal lobe epilepsy: from patient A to Doctor Z. *Epilepsy and Behavior*, **2006**, 9, 524-31	3.2	4
19	Child Neurology: muscular dystrophy without contractures. *Neurology*, **2017**, 88, e199-e203	6.5	3
18	ACTA1-myopathy with prominent finger flexor weakness and rimmed vacuoles. *Neuromuscular Disorders*, **2019**, 29, 388-391	2.9	3
17	Schwann cell heterogeneity--might it underlie the diversity seen in Schwann cell tumors?. *Human Pathology*, **1994**, 25, 1113	3.7	3
16	Differential metabolism of hydroxyeicosatetraenoic acid isomers by mouse cerebromicrovascular endothelium. *Journal of Neurochemistry*, **1992**, 58, 374-82	6	3
15	Primary germinoma of the spinal cord: a case report with 28-year follow-up and review of the literature **1995**, 90, 657		3
14	Congenital myasthenic syndrome caused by a frameshift insertion mutation in. *Neurology: Genetics*, **2020**, 6, e468	3.8	3
13	Pathogenic variants in TNNC2 cause congenital myopathy due to an impaired force response to calcium. *Journal of Clinical Investigation*, **2021**, 131,	15.9	3
12	Validation of Optical Genome Mapping for the Molecular Diagnosis of Facioscapulohumeral Muscular Dystrophy. *Journal of Molecular Diagnostics*, **2021**, 23, 1506-1514	5.1	3
11	Dystrophinopathy muscle biopsies in the genetic testing ERA: One center's data. *Muscle and Nerve*, **2018**, 58, 149	3.4	2
10	Clinical Reasoning: A 30-year-old man with progressive weakness and atrophy. *Neurology*, **2016**, 87, e227-e230	6.5	2
9	A novel noncoding mutation in early onset limb-girdle muscular dystrophy. *Neurology: Genetics*, **2020**, 6, e388	3.8	1
8	Thalamic ependymoma presenting as recurrent subarachnoid hemorrhage. *Journal of Stroke and Cerebrovascular Diseases*, **1992**, 2, 106-9	2.8	1
7	The inflammatory pathology of dysferlinopathy is distinct from calpainopathy, Becker muscular dystrophy, and inflammatory myopathies.. *Acta Neuropathologica Communications*, **2022**, 10, 17	7.3	1

List of Publications

6	Cobblestone Malformation in LAMA2 Congenital Muscular Dystrophy (MDC1A). *Journal of Neuropathology and Experimental Neurology*, **2020**, 79, 998-1010	3.1	1
5	Refractory infantile high-grade glioma containing TRK-fusion responds to larotrectinib. *Pediatric Blood and Cancer*, **2021**, 68, e28868	3	0
4	Eicosanoid Production by Isolated Cerebral Microvessels and Cultured Cerebral Endothelium. *Annals of the New York Academy of Sciences*, **1989**, 559, 471-473	6.5	
3	Danon disease (autophagic vacuolar myopathy): clinicopathologic features of a kindred. *FASEB Journal*, **2007**, 21, A399	0.9	
2	Common mutations detected for diagnosis of limb-girdle muscular dystrophy. *FASEB Journal*, **2007**, 21, A398	0.9	
1	Congenital Myasthenic Syndromes **2021**, 313-316		