

# Mark Andrew Tarnopolsky

## List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

356  
papers

16,573  
citations

72  
h-index

117  
g-index

369  
ext. papers

19,073  
ext. citations

4.2  
avg, IF

6.77  
L-index

#	Paper	IF	Citations
356	Caffeine blocks SREBP2-induced hepatic PCSK9 expression to enhance LDLR-mediated cholesterol clearance.. <i>Nature Communications</i> , <b>2022</b> , 13, 770	17.4	5
355	Possible association between rhabdomyolysis and mRNA SARS-CoV-2 vaccination in a patient with gene mutation.. <i>Cmaj</i> , <b>2022</b> , 194, E252-E256	3.5	0
354	Aerobic exercise elicits clinical adaptations in myotonic dystrophy type 1 patients independent of pathophysiological changes.. <i>Journal of Clinical Investigation</i> , <b>2022</b> ,	15.9	2
353	Life-long exercise training and inherited aerobic endurance capacity produce converging gut microbiome signatures in rodents.. <i>Physiological Reports</i> , <b>2022</b> , 10, e15215	2.6	3
352	Functional characterization of variants of unknown significance in a spinocerebellar ataxia patient using an unsupervised machine learning pipeline.. <i>Human Genome Variation</i> , <b>2022</b> , 9, 10	1.8	0
351	Genetic, structural and clinical analysis of spastic paraplegia 4.. <i>Parkinsonism and Related Disorders</i> , <b>2022</b> , 98, 62-69	3.6	0
350	Multi-Ingredient Supplement Supports Mitochondrial Health through Interleukin-15 Signaling in Older Adult Human Dermal Fibroblasts. <i>Cosmetics</i> , <b>2022</b> , 9, 47	2.7	
349	Heterozygous frameshift variants in HNRNPA2B1 cause early-onset oculopharyngeal muscular dystrophy.. <i>Nature Communications</i> , <b>2022</b> , 13, 2306	17.4	1
348	Validation and clinical performance of a combined nuclear-mitochondrial next-generation sequencing and copy number variant analysis panel in a Canadian population. <i>American Journal of Medical Genetics, Part A</i> , <b>2021</b> , 185, 486-499	2.5	2
347	Immune-mediated Necrotizing Myopathy following BNT162b2 Vaccination in a Patient with Antibodies against Receptor-binding Domain of SARS-CoV-2 and Signal Recognition Particle.. <i>Muscle and Nerve</i> , <b>2021</b> ,	3.4	0
346	Effects of an acute exercise bout in hypoxia on extracellular vesicle release in healthy and prediabetic subjects.. <i>American Journal of Physiology - Regulatory Integrative and Comparative Physiology</i> , <b>2021</b> ,	3.2	3
345	A Novel Multi-Ingredient Supplement Activates a Browning Program in White Adipose Tissue and Mitigates Weight Gain in High-Fat Diet-Fed Mice. <i>Nutrients</i> , <b>2021</b> , 13,	6.7	2
344	Ontario Newborn Screening for Spinal Muscular Atrophy: The First Year. <i>Canadian Journal of Neurological Sciences</i> , <b>2021</b> , 1-7	1	
343	Newborn Screening for Spinal Muscular Atrophy: Ontario Testing and Follow-up Recommendations. <i>Canadian Journal of Neurological Sciences</i> , <b>2021</b> , 48, 504-511	1	4
342	Human skeletal muscle fiber type-specific responses to sprint interval and moderate-intensity continuous exercise: acute and training-induced changes. <i>Journal of Applied Physiology</i> , <b>2021</b> , 130, 1001-1014	3.7	5
341	Impaired Function and Altered Morphology in the Skeletal Muscles of Adult Men and Women With Type 1 Diabetes. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2021</b> , 106, 2405-2422	5.6	2
340	Childhood amyotrophic lateral sclerosis caused by excess sphingolipid synthesis. <i>Nature Medicine</i> , <b>2021</b> , 27, 1197-1204	50.5	22

339	Management of mitochondrial diabetes in the era of novel therapies. <i>Journal of Diabetes and Its Complications</i> , <b>2021</b> , 35, 107584	3.2	11
338	Comprehensive genetic sequence and copy number analysis for Charcot-Marie-Tooth disease in a Canadian cohort of 2517 patients. <i>Journal of Medical Genetics</i> , <b>2021</b> , 58, 284-288	5.8	2
337	A biallelic pathogenic variant in the OGDH gene results in a neurological disorder with features of a mitochondrial disease. <i>Journal of Inherited Metabolic Disease</i> , <b>2021</b> , 44, 388-400	5.4	9
336	Mitochondrial neuropathy and neurogenic features in mitochondrial myopathy. <i>Mitochondrion</i> , <b>2021</b> , 56, 52-61	4.9	5
335	An autosomal dominant neurological disorder caused by de novo variants in FAR1 resulting in uncontrolled synthesis of ether lipids. <i>Genetics in Medicine</i> , <b>2021</b> , 23, 740-750	8.1	7
334	Acute, Exercise-Induced Alterations in Cytokines and Chemokines in the Blood Distinguish Physically Active and Sedentary Aging. <i>Journals of Gerontology - Series A Biological Sciences and Medical Sciences</i> , <b>2021</b> , 76, 811-818	6.4	0
333	Variants in GNAI1 cause a syndrome associated with variable features including developmental delay, seizures, and hypotonia. <i>Genetics in Medicine</i> , <b>2021</b> , 23, 881-887	8.1	1
332	A pathogenic UFSP2 variant in an autosomal recessive form of pediatric neurodevelopmental anomalies and epilepsy. <i>Genetics in Medicine</i> , <b>2021</b> , 23, 900-908	8.1	2
331	POLRMT mutations impair mitochondrial transcription causing neurological disease. <i>Nature Communications</i> , <b>2021</b> , 12, 1135	17.4	3
330	Evidence for Non-Mendelian Inheritance in Spastic Paraplegia 7. <i>Movement Disorders</i> , <b>2021</b> , 36, 1664-1675		7
329	Normal to enhanced intrinsic mitochondrial respiration in skeletal muscle of middle- to older-aged women and men with uncomplicated type 1 diabetes. <i>Diabetologia</i> , <b>2021</b> , 64, 2517-2533	10.3	0
328	Alterations in skeletal muscle repair in young adults with type 1 diabetes mellitus. <i>American Journal of Physiology - Cell Physiology</i> , <b>2021</b> , 321, C876-C883	5.4	0
327	ABHD16A deficiency causes a complicated form of hereditary spastic paraplegia associated with intellectual disability and cerebral anomalies. <i>American Journal of Human Genetics</i> , <b>2021</b> , 108, 2017-2023	11	1
326	Dual molecular diagnoses in a neurometabolic specialty clinic. <i>American Journal of Medical Genetics, Part A</i> , <b>2021</b> , 185, 766-773	2.5	3
325	A mitochondrial disorder with ptosis and exercise intolerance without ophthalmoparesis secondary to m.5865T>C variant. <i>Mitochondrion</i> , <b>2020</b> , 53, 150-153	4.9	1
324	Expanding Clinical Presentations Due to Variations in THOC2 mRNA Nuclear Export Factor. <i>Frontiers in Molecular Neuroscience</i> , <b>2020</b> , 13, 12	6.1	4
323	Response to "Relation between intra-mitochondrial inclusions and pathophysiology of mitochondrial myopathy remains unprecise". <i>Journal of the Neurological Sciences</i> , <b>2020</b> , 414, 116895	3.2	
322	Intracellular calcium leak as a therapeutic target for RYR1-related myopathies. <i>Acta Neuropathologica</i> , <b>2020</b> , 139, 1089-1104	14.3	9

321	Increased intra-mitochondrial lipofuscin aggregates with spherical dense body formation in mitochondrial myopathy. <i>Journal of the Neurological Sciences</i> , <b>2020</b> , 413, 116816	3.2	6
320	Congenital myasthenic syndrome-associated agrin variants affect clustering of acetylcholine receptors in a domain-specific manner. <i>JCI Insight</i> , <b>2020</b> , 5,	9.9	6
319	Lessons of the month: A breathless severe asthmatic in the genomic era: Occam's razor or Hickam's dictum?. <i>Clinical Medicine</i> , <b>2020</b> , 20, e264-e266	1.9	1
318	Expanding the Phenotype: Neurodevelopmental Disorder, Mitochondrial, With Abnormal Movements and Lactic Acidosis, With or Without Seizures (NEMMLAS) Due to WARS2 Biallelic Variants, Encoding Mitochondrial Tryptophanyl-tRNA Synthase. <i>Journal of Child Neurology</i> , <b>2020</b> , 35, 176-177	2.5	
317	Choline transporter-like 1 deficiency causes a new type of childhood-onset neurodegeneration. <i>Brain</i> , <b>2020</b> , 143, 94-111	11.2	7
316	Muscle and serum myostatin expression in type 1 diabetes. <i>Physiological Reports</i> , <b>2020</b> , 8, e14500	2.6	6
315	Assessing non-Mendelian inheritance in inherited axonopathies. <i>Genetics in Medicine</i> , <b>2020</b> , 22, 2114-2119	9.1	7
314	Bone marrow-derived mitochondrial DNA has limited capacity for inter-tissue transfer in vivo. <i>FASEB Journal</i> , <b>2020</b> , 34, 9297-9306	0.9	2
313	A Five-Ingredient Nutritional Supplement and Home-Based Resistance Exercise Improve Lean Mass and Strength in Free-Living Elderly. <i>Nutrients</i> , <b>2020</b> , 12,	6.7	16
312	Severe chorioretinal atrophy in Boucher-Neuhauser syndrome. <i>Canadian Journal of Ophthalmology</i> , <b>2020</b> , 55, e26-e28	1.4	4
311	Neurodevelopmental and associated changes in a patient with Xp22.31 duplication. <i>Neurological Sciences</i> , <b>2020</b> , 41, 713-716	3.5	1
310	Nutrition for Special Populations: Young, Female, and Masters Athletes. <i>International Journal of Sport Nutrition and Exercise Metabolism</i> , <b>2019</b> , 29, 220-227	4.4	26
309	Statin administration activates system xC in skeletal muscle: a potential mechanism explaining statin-induced muscle pain. <i>American Journal of Physiology - Cell Physiology</i> , <b>2019</b> , 317, C894-C899	5.4	3
308	Genotypes of chronic progressive external ophthalmoplegia in a large adult-onset cohort. <i>Mitochondrion</i> , <b>2019</b> , 49, 227-231	4.9	13
307	Diagnosis of 'possible' mitochondrial disease: an existential crisis. <i>Journal of Medical Genetics</i> , <b>2019</b> , 56, 123-130	5.8	27
306	Lifelong aerobic exercise protects against inflammaging and cancer. <i>PLoS ONE</i> , <b>2019</b> , 14, e0210863	3.7	37
305	Paraspinal muscle ladybird homeobox 1 (LBX1) in adolescent idiopathic scoliosis: a cross-sectional study. <i>Spine Journal</i> , <b>2019</b> , 19, 1911-1916	4	6
304	Mitochondria and Aging-The Role of Exercise as a Countermeasure. <i>Biology</i> , <b>2019</b> , 8,	4.9	26

303	Myasthenia graves-like symptoms associated with rare mitochondrial mutation (m.5728T>C). <i>Mitochondrion</i> , <b>2019</b> , 47, 139-140	4.9	3
302	Neurogenic Muscle Biopsy Findings Are Common in Mitochondrial Myopathy. <i>Journal of Neuropathology and Experimental Neurology</i> , <b>2019</b> , 78, 508-514	3.1	5
301	Diagnostic Utility of Genome-wide DNA Methylation Testing in Genetically Unsolved Individuals with Suspected Hereditary Conditions. <i>American Journal of Human Genetics</i> , <b>2019</b> , 104, 685-700	11	57
300	Truncating Mutations in UBAP1 Cause Hereditary Spastic Paraplegia. <i>American Journal of Human Genetics</i> , <b>2019</b> , 104, 767-773	11	20
299	Obesity and muscle-macrophage crosstalk in humans and mice: A systematic review. <i>Obesity Reviews</i> , <b>2019</b> , 20, 1572-1596	10.6	4
298	Expanding the Phenotype: Neurodevelopmental Disorder, Mitochondrial, With Abnormal Movements and Lactic Acidosis, With or Without Seizures (NEMMLAS) due to WARS2 Biallelic Variants, Encoding Mitochondrial Tryptophanyl-tRNA Synthase. <i>Journal of Child Neurology</i> , <b>2019</b> , 34, 776-784	2.5	6
297	Muscle problems in juvenile-onset acid maltase deficiency (Pompe disease). <i>Paediatrics and Child Health</i> , <b>2019</b> , 24, 270-271	0.7	1
296	Expanding the Clinical Spectrum of -Related Mitochondrial Cytopathy. <i>Frontiers in Neurology</i> , <b>2019</b> , 10, 981	4.1	8
295	Effect of short-term, high-intensity exercise training on human skeletal muscle citrate synthase maximal activity: single versus multiple bouts per session. <i>Applied Physiology, Nutrition and Metabolism</i> , <b>2019</b> , 44, 1391-1394	3	0
294	Loss of the sphingolipid desaturase DEGS1 causes hypomyelinating leukodystrophy. <i>Journal of Clinical Investigation</i> , <b>2019</b> , 129, 1240-1256	15.9	37
293	Nutrition and exercise in Pompe disease. <i>Annals of Translational Medicine</i> , <b>2019</b> , 7, 282	3.2	4
292	Expanding the Boundaries of RNA Sequencing as a Diagnostic Tool for Rare Mendelian Disease. <i>American Journal of Human Genetics</i> , <b>2019</b> , 104, 466-483	11	110
291	An evaluation of genetic causes and environmental risks for bilateral optic atrophy. <i>PLoS ONE</i> , <b>2019</b> , 14, e0225656	3.7	3
290	Clinical features related to statin-associated muscle symptoms. <i>Muscle and Nerve</i> , <b>2019</b> , 59, 537-543	3.4	7
289	Complete elimination of a pathogenic homoplasmic mtDNA mutation in one generation. <i>Mitochondrion</i> , <b>2019</b> , 45, 18-21	4.9	1
288	CPEO - Like mitochondrial myopathy associated with m.8340G>A mutation. <i>Mitochondrion</i> , <b>2019</b> , 46, 69-72	4.9	3
287	Clinical and demographic features of chronic progressive external ophthalmoplegia in a large adult-onset cohort. <i>Mitochondrion</i> , <b>2019</b> , 44, 15-19	4.9	13
286	An evaluation of genetic causes and environmental risks for bilateral optic atrophy <b>2019</b> , 14, e0225656		

285	An evaluation of genetic causes and environmental risks for bilateral optic atrophy <b>2019</b> , 14, e0225656		
284	An evaluation of genetic causes and environmental risks for bilateral optic atrophy <b>2019</b> , 14, e0225656		
283	An evaluation of genetic causes and environmental risks for bilateral optic atrophy <b>2019</b> , 14, e0225656		
282	Skeletal muscle fiber-type-specific changes in markers of capillary and mitochondrial content after low-volume interval training in overweight women. <i>Physiological Reports</i> , <b>2018</b> , 6, e13597	2.6	19
281	The prognosis for glycemic status among children and youth with obesity 2 years after entering a weight management program. <i>Pediatric Diabetes</i> , <b>2018</b> , 19, 874-881	3.6	3
280	Altered mitochondrial bioenergetics and ultrastructure in the skeletal muscle of young adults with type 1 diabetes. <i>Diabetologia</i> , <b>2018</b> , 61, 1411-1423	10.3	46
279	Next-Generation Sequencing to Diagnose Muscular Dystrophy, Rhabdomyolysis, and HyperCKemia. <i>Canadian Journal of Neurological Sciences</i> , <b>2018</b> , 45, 262-268	1	16
278	POLR3A variants in hereditary spastic paraplegia and ataxia. <i>Brain</i> , <b>2018</b> , 141, e1	11.2	14
277	Exosomes as Mediators of the Systemic Adaptations to Endurance Exercise. <i>Cold Spring Harbor Perspectives in Medicine</i> , <b>2018</b> , 8,	5.4	77
276	Blunted satellite cell response is associated with dysregulated IGF-1 expression after exercise with age. <i>European Journal of Applied Physiology</i> , <b>2018</b> , 118, 2225-2231	3.4	5
275	Aberrant Drp1-mediated mitochondrial division presents in humans with variable outcomes. <i>Human Molecular Genetics</i> , <b>2018</b> , 27, 3710-3719	5.6	21
274	Severe neurocognitive and growth disorders due to variation in THOC2, an essential component of nuclear mRNA export machinery. <i>Human Mutation</i> , <b>2018</b> , 39, 1126-1138	4.7	8
273	Cardiopulmonary Exercise Testing Reflects Improved Exercise Capacity in Response to Treatment in Morquio A Patients: Results of a 52-Week Pilot Study of Two Different Doses of Elosulfase Alfa. <i>JIMD Reports</i> , <b>2018</b> , 42, 9-17	1.9	4
272	The genotypic and phenotypic spectrum of MTO1 deficiency. <i>Molecular Genetics and Metabolism</i> , <b>2018</b> , 123, 28-42	3.7	18
271	BAFopathies' DNA methylation epi-signatures demonstrate diagnostic utility and functional continuum of Coffin-Siris and Nicolaides-Baraitser syndromes. <i>Nature Communications</i> , <b>2018</b> , 9, 4885	17.4	48
270	Myopathies Related to Glycogen Metabolism Disorders. <i>Neurotherapeutics</i> , <b>2018</b> , 15, 915-927	6.4	21
269	Novel Association of a De Novo CALM2 Mutation With Long QT Syndrome and Hypertrophic Cardiomyopathy. <i>Circulation Genomic and Precision Medicine</i> , <b>2018</b> , 11, e002255	5.2	5
268	Molecular characterization of NRXN1 deletions from 19,263 clinical microarray cases identifies exons important for neurodevelopmental disease expression. <i>Genetics in Medicine</i> , <b>2017</b> , 19, 53-61	8.1	43

267	Effect of sex on the acute skeletal muscle response to sprint interval exercise. <i>Experimental Physiology</i> , <b>2017</b> , 102, 354-365	2.4	19
266	Perspectives on Exertional Rhabdomyolysis. <i>Sports Medicine</i> , <b>2017</b> , 47, 33-49	10.6	65
265	encephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. <i>Journal of Medical Genetics</i> , <b>2017</b> , 54, 460-470	5.8	109
264	A mutation in the TMEM65 gene results in mitochondrial myopathy with severe neurological manifestations. <i>European Journal of Human Genetics</i> , <b>2017</b> , 25, 744-751	5.3	8
263	mutations are associated with features of complex hereditary spastic paraparesis. <i>Neurology</i> , <b>2017</b> , 89, 2210-2211	6.5	11
262	Higher oxidative stress in skeletal muscle of McArdle disease patients. <i>Molecular Genetics and Metabolism Reports</i> , <b>2017</b> , 12, 69-75	1.8	8
261	Patient care standards for primary mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. <i>Genetics in Medicine</i> , <b>2017</b> , 19,	8.1	113
260	Clinical Manifestations Associated With the N-Terminal-Acetyltransferase NAA10 Gene Mutation in a Girl: Ogden Syndrome. <i>Pediatric Neurology</i> , <b>2017</b> , 76, 82-85	2.9	15
259	Response to Newman et al. <i>Genetics in Medicine</i> , <b>2017</b> , 19,	8.1	2
258	Optimizing the methodology for measuring supraclavicular skin temperature using infrared thermography; implications for measuring brown adipose tissue activity in humans. <i>Scientific Reports</i> , <b>2017</b> , 7, 11934	4.9	15
257	Spinocerebellar ataxia type 29 due to mutations in ITPR1: a case series and review of this emerging congenital ataxia. <i>Orphanet Journal of Rare Diseases</i> , <b>2017</b> , 12, 121	4.2	27
256	Males With MECP2 C-terminal-Related Atypical Rett Syndromes and Their Carrier Mothers. <i>Pediatric Neurology</i> , <b>2017</b> , 67, 98-101	2.9	1
255	Myostatin inhibitor ACE-031 treatment of ambulatory boys with Duchenne muscular dystrophy: Results of a randomized, placebo-controlled clinical trial. <i>Muscle and Nerve</i> , <b>2017</b> , 55, 458-464	3.4	124
254	Superior mitochondrial adaptations in human skeletal muscle after interval compared to continuous single-leg cycling matched for total work. <i>Journal of Physiology</i> , <b>2017</b> , 595, 2955-2968	3.9	105
253	Complex IV <b>2016</b> , 279-285		
252	Salsalate (Salicylate) Uncouples Mitochondria, Improves Glucose Homeostasis, and Reduces Liver Lipids Independent of AMPK- $\beta$ . <i>Diabetes</i> , <b>2016</b> , 65, 3352-3361	0.9	41
251	Digital PCR methods improve detection sensitivity and measurement precision of low abundance mtDNA deletions. <i>Scientific Reports</i> , <b>2016</b> , 6, 25186	4.9	43
250	Decreased Satellite Cell Number and Function in Humans and Mice With Type 1 Diabetes Is the Result of Altered Notch Signaling. <i>Diabetes</i> , <b>2016</b> , 65, 3053-61	0.9	26

249	Solid organ transplantation in primary mitochondrial disease: Proceed with caution. <i>Molecular Genetics and Metabolism</i> , <b>2016</b> , 118, 178-184	3.7	40
248	Exercise-induced mitochondrial p53 repairs mtDNA mutations in mutator mice. <i>Skeletal Muscle</i> , <b>2016</b> , 6, 7	5.1	53
247	Metabolic Myopathies. <i>CONTINUUM Lifelong Learning in Neurology</i> , <b>2016</b> , 22, 1829-1851	3	18
246	Complex V Disorders <b>2016</b> , 287-291		
245	Chronic Progressive External Ophthalmoplegia (CPEO) <b>2016</b> , 49-53		
244	Complex I Deficiency <b>2016</b> , 257-264		
243	Twelve Weeks of Sprint Interval Training Improves Indices of Cardiometabolic Health Similar to Traditional Endurance Training despite a Five-Fold Lower Exercise Volume and Time Commitment. <i>PLoS ONE</i> , <b>2016</b> , 11, e0154075	3.7	177
242	Targeted Re-Sequencing Emulsion PCR Panel for Myopathies: Results in 94 Cases. <i>Journal of Neuromuscular Diseases</i> , <b>2016</b> , 3, 209-225	5	13
241	Genetic Myopathies Initially Diagnosed and Treated as Inflammatory Myopathy. <i>Canadian Journal of Neurological Sciences</i> , <b>2016</b> , 43, 381-4	1	5
240	The potential of endurance exercise-derived exosomes to treat metabolic diseases. <i>Nature Reviews Endocrinology</i> , <b>2016</b> , 12, 504-17	15.2	191
239	De novo mutations in CSNK2A1 are associated with neurodevelopmental abnormalities and dysmorphic features. <i>Human Genetics</i> , <b>2016</b> , 135, 699-705	6.3	32
238	Pompe Disease: Diagnosis and Management. Evidence-Based Guidelines from a Canadian Expert Panel. <i>Canadian Journal of Neurological Sciences</i> , <b>2016</b> , 43, 472-85	1	37
237	Two novel mitochondrial tRNA mutations, A7495G (tRNA) and C5577T (tRNA), are associated with seizures and cardiac dysfunction. <i>Mitochondrion</i> , <b>2016</b> , 31, 40-44	4.9	4
236	Effects of age and unaccustomed resistance exercise on mitochondrial transcript and protein abundance in skeletal muscle of men. <i>American Journal of Physiology - Regulatory Integrative and Comparative Physiology</i> , <b>2015</b> , 308, R734-41	3.2	27
235	Exome sequencing identifies complex I NDUFV2 mutations as a novel cause of Leigh syndrome. <i>European Journal of Paediatric Neurology</i> , <b>2015</b> , 19, 525-32	3.8	22
234	Metabolomic analysis of exercise effects in the POLG mitochondrial DNA mutator mouse brain. <i>Neurobiology of Aging</i> , <b>2015</b> , 36, 2972-2983	5.6	23
233	Prenatal growth restriction, retinal dystrophy, diabetes insipidus and white matter disease: expanding the spectrum of PRPS1-related disorders. <i>European Journal of Human Genetics</i> , <b>2015</b> , 23, 310-6	5.3	23
232	Phospholamban overexpression in mice causes a centronuclear myopathy-like phenotype. <i>DMM Disease Models and Mechanisms</i> , <b>2015</b> , 8, 999-1009	4.1	21



231	Satellite cell activity, without expansion, after nonhypertrophic stimuli. <i>American Journal of Physiology - Regulatory Integrative and Comparative Physiology</i> , <b>2015</b> , 309, R1101-11	3.2	38
230	Dysfunctional mitochondria? Let's make more exercise in a bottle. <i>Mitochondrion</i> , <b>2015</b> , 24, S4	4.9	
229	Two cases of congenital myasthenic syndrome with vocal cord paralysis. <i>Neurology</i> , <b>2015</b> , 84, 1281-2	6.5	16
228	Association of depression & health related quality of life with body composition in children and youth with obesity. <i>Journal of Affective Disorders</i> , <b>2015</b> , 172, 18-23	6.6	102
227	Sex differences in skeletal muscle phosphatase and tensin homolog deleted on chromosome 10 (PTEN) levels: a cross-sectional study. <i>Scientific Reports</i> , <b>2015</b> , 5, 9154	4.9	8
226	Adiposity and immune-muscle crosstalk in South Asians & Europeans: A cross-sectional study. <i>Scientific Reports</i> , <b>2015</b> , 5, 14521	4.9	4
225	MG-123 Exonic and intronic NRXN1 deletions: Novel genotype-phenotype correlations. <i>Journal of Medical Genetics</i> , <b>2015</b> , 52, A9.1-A9	5.8	
224	Exercise-stimulated interleukin-15 is controlled by AMPK and regulates skin metabolism and aging. <i>Aging Cell</i> , <b>2015</b> , 14, 625-34	9.9	75
223	Sodium bicarbonate ingestion augments the increase in PGC-1 $\alpha$ mRNA expression during recovery from intense interval exercise in human skeletal muscle. <i>Journal of Applied Physiology</i> , <b>2015</b> , 119, 1303-12	3.7	30
222	Feasibility and Reliability of Muscle Strength Testing in Critically Ill Children. <i>Journal of Pediatric Intensive Care</i> , <b>2015</b> , 4, 218-224	1	8
221	Diagnosis and management of mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. <i>Genetics in Medicine</i> , <b>2015</b> , 17, 689-701	8.1	284
220	Polymerase gamma mutator mice rely on increased glycolytic flux for energy production. <i>Mitochondrion</i> , <b>2015</b> , 21, 19-26	4.9	12
219	Exercise-Induced Amelioration of Diet-Induced Obesity and Diabetes is Not Regulated by Irisin. <i>FASEB Journal</i> , <b>2015</b> , 29, 992.4	0.9	
218	Intermittent and continuous high-intensity exercise training induce similar acute but different chronic muscle adaptations. <i>Experimental Physiology</i> , <b>2014</b> , 99, 782-91	2.4	74
217	Fluvastatin causes NLRP3 inflammasome-mediated adipose insulin resistance. <i>Diabetes</i> , <b>2014</b> , 63, 3742-7	6.9	86
216	Exercise as a therapeutic strategy for primary mitochondrial cytopathies. <i>Journal of Child Neurology</i> , <b>2014</b> , 29, 1225-34	2.5	26
215	Functional impairment in patients with sporadic Inclusion Body Myositis. <i>Canadian Journal of Neurological Sciences</i> , <b>2014</b> , 41, 253-9	1	2
214	Statin-associated Autoimmune Myopathies: A Pathophysiologic Spectrum. <i>Canadian Journal of Neurological Sciences</i> , <b>2014</b> , 41, 638-47	1	19

213	Defects in mitochondrial DNA replication and oxidative damage in muscle of mtDNA mutator mice. <i>Free Radical Biology and Medicine</i> , <b>2014</b> , 75, 241-51	7.8	39
212	The unfolded protein response is triggered following a single, unaccustomed resistance-exercise bout. <i>American Journal of Physiology - Regulatory Integrative and Comparative Physiology</i> , <b>2014</b> , 307, R664-9	3.2	43
211	Genome-wide DNA methylation changes with age in disease-free human skeletal muscle. <i>Aging Cell</i> , <b>2014</b> , 13, 360-6	9.9	110
210	Evidence for synergistic effects of PRNP and ATP7B mutations in severe neuropsychiatric deterioration. <i>BMC Medical Genetics</i> , <b>2014</b> , 15, 22	2.1	9
209	Impact of habitual exercise on the strength of individuals with myotonic dystrophy type 1. <i>American Journal of Physical Medicine and Rehabilitation</i> , <b>2014</b> , 93, 739-46; quiz 747-8	2.6	16
208	Three minutes of all-out intermittent exercise per week increases skeletal muscle oxidative capacity and improves cardiometabolic health. <i>PLoS ONE</i> , <b>2014</b> , 9, e111489	3.7	107
207	Mitochondrial encephalopathy with lactic acidosis and stroke-like episodes (MELAS) may respond to adjunctive ketogenic diet. <i>Pediatric Neurology</i> , <b>2014</b> , 50, 498-502	2.9	56
206	Redox state and mitochondrial respiratory chain function in skeletal muscle of LGMD2A patients. <i>PLoS ONE</i> , <b>2014</b> , 9, e102549	3.7	14
205	The order of exercise during concurrent training for rehabilitation does not alter acute genetic expression, mitochondrial enzyme activity or improvements in muscle function. <i>PLoS ONE</i> , <b>2014</b> , 9, e109189	3.7	18
204	Dysferlin aggregation in limb-girdle muscular dystrophy type 2B/Miyoshi Myopathy necessitates mutational screen for diagnosis [corrected]. <i>Muscle and Nerve</i> , <b>2013</b> , 47, 740-7	3.4	14
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202	Endurance training modulates intramyocellular lipid compartmentalization and morphology in skeletal muscle of lean and obese women. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2013</b> , 98, 4852-62	5.6	55
201	Oxidative stress and Nrf2 signaling in McArdle disease. <i>Molecular Genetics and Metabolism</i> , <b>2013</b> , 110, 297-302	3.7	20
200	Monocarboxylate transporters and mitochondrial creatine kinase protein content in McArdle disease. <i>Molecular Genetics and Metabolism</i> , <b>2013</b> , 108, 259-62	3.7	6
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198	Leigh syndrome associated with mitochondrial complex I deficiency due to novel mutations in NDUFV1 and NDUFS2. <i>Gene</i> , <b>2013</b> , 516, 162-7	3.8	32
197	Severe infantile Leigh syndrome associated with a rare mitochondrial ND6 mutation, m.14487T>C. <i>American Journal of Medical Genetics, Part A</i> , <b>2013</b> , 161A, 2020-3	2.5	17
196	Metabolite measurements in the caudate nucleus, anterior cingulate cortex and hippocampus among patients with mitochondrial disorders: a case-control study using proton magnetic resonance spectroscopy. <i>CMAJ Open</i> , <b>2013</b> , 1, E48-55	2.5	5

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194	Eccentric exercise increases satellite cell content in type II muscle fibers. <i>Medicine and Science in Sports and Exercise</i> , <b>2013</b> , 45, 230-7	1.2	65
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192	Creatine for treating muscle disorders. <i>The Cochrane Library</i> , <b>2013</b> , CD004760	5.2	39
191	Substrate Utilization in Female Athletes <b>2013</b> , 1-24		1
190	Elevated mitochondrial oxidative stress impairs metabolic adaptations to exercise in skeletal muscle. <i>PLoS ONE</i> , <b>2013</b> , 8, e81879	3.7	18
189	Supplementation with lipoic acid, CoQ10, and vitamin E augments running performance and mitochondrial function in female mice. <i>PLoS ONE</i> , <b>2013</b> , 8, e60722	3.7	30
188	Markers of skeletal muscle mitochondrial function and lipid accumulation are moderately associated with the homeostasis model assessment index of insulin resistance in obese men. <i>PLoS ONE</i> , <b>2013</b> , 8, e66322	3.7	34
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178	Effects of creatine and exercise on skeletal muscle of FRG1-transgenic mice. <i>Canadian Journal of Neurological Sciences</i> , <b>2012</b> , 39, 225-31	1	5

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149	Effects of exercise and corticotrophin-releasing factor 2 receptor agonist on skeletal muscle of mdx mice. <i>FASEB Journal</i> , <b>2010</b> , 24, 806.13	0.9	
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54	Novel SCO2 mutation (G1521A) presenting as a spinal muscular atrophy type I phenotype. <i>American Journal of Medical Genetics Part A</i> , <b>2004</b> , 125A, 310-4		39
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