Miguel Angel MartÃ-n

List of Publications by Year
in descending order

207	7,254	66343	71685
papers	citations	42 h-index	76 g-index

213	213	213	8741
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Elevated glutamate and decreased glutamine levels in the cerebrospinal fluid of patients with MELAS syndrome. Journal of Neurology, 2022, 269, 3238-3248.	3.6	7
2	Apoptosis-Inducing Factor Deficiency Induces Tissue-Specific Alterations in Autophagy: Insights from a Preclinical Model of Mitochondrial Disease and Exercise Training Effects. Antioxidants, 2022, 11, 510.	5.1	0
3	Long-Term Exercise Intervention in Patients with McArdle Disease: Clinical and Aerobic Fitness Benefits. Medicine and Science in Sports and Exercise, 2022, 54, 1231-1241.	0.4	7
4	A novel TRMT5 mutation causes a complex inherited neuropathy syndrome: The role of nerve pathology in defining a demyelinating neuropathy. Neuropathology and Applied Neurobiology, 2022, 48, e12817.	3.2	1
5	Muscle MRI characteristic pattern for late-onset TK2 deficiency diagnosis. Journal of Neurology, 2022, 269, 3550-3562.	3.6	4
6	Multicentric Standardization of Protocols for the Diagnosis of Human Mitochondrial Respiratory Chain Defects. Antioxidants, 2022, 11, 741.	5.1	4
7	Clinical, Histological, and Genetic Features of 25 Patients with Autosomal Dominant Progressive External Ophthalmoplegia (ad-PEO)/PEO-Plus Due to TWNK Mutations. Journal of Clinical Medicine, 2022, 11, 22.	2.4	5
8	Preclinical Research in McArdle Disease: A Review of Research Models and Therapeutic Strategies. Genes, 2022, 13, 74.	2.4	4
9	Identification of Potential Muscle Biomarkers in McArdle Disease: Insights from Muscle Proteome Analysis. International Journal of Molecular Sciences, 2022, 23, 4650.	4.1	0
10	Metrics of Progression and Prognosis in Untreated Adults With Thymidine Kinase 2 Deficiency: An Observational Study. Neuromuscular Disorders, 2022, , .	0.6	2
11	Small GTPases of the Ras superfamily and glycogen phosphorylase regulation in T cells. Small GTPases, 2021, 12, 106-113.	1.6	10
12	Soluble fms-like tyrosine kinase-1: a potential early predictor of respiratory failure in COVID-19 patients. Clinical Chemistry and Laboratory Medicine, 2021, 59, e289-e292.	2.3	4
13	Recurrent rhabdomyolysis and exercise intolerance: A new phenotype of late-onset thymidine kinase 2 deficiency. Molecular Genetics and Metabolism Reports, 2021, 26, 100701.	1.1	6
14	Pearls & Oy-sters: Hickam's Dictum in Genetic Myopathies. Neurology, 2021, 96, 1007-1009.	1.1	0
15	Plasma LDH: A specific biomarker for lung affectation in COVID-19?. Practical Laboratory Medicine, 2021, 25, e00226.	1.3	10
16	Preferent Diaphragmatic Involvement in TK2 Deficiency: An Autopsy Case Study. International Journal of Molecular Sciences, 2021, 22, 5598.	4.1	4
17	Plasma Gelsolin Reinforces the Diagnostic Value of FGF-21 and GDF-15 for Mitochondrial Disorders. International Journal of Molecular Sciences, 2021, 22, 6396.	4.1	8
18	Pathogenetic and Prognostic Implications of Increased Mitochondrial Content in Multiple Myeloma. Cancers, 2021, 13, 3189.	3.7	3

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19	Biallelic variants in genes previously associated with dominant inheritance: CACNA1A, RET and SLC20A2. European Journal of Human Genetics, 2021, 29, 1520-1526.	2.8	10
20	SILAC-based complexome profiling dissects the structural organization of the human respiratory supercomplexes in SCAFIKO cells. Biochimica Et Biophysica Acta - Bioenergetics, 2021, 1862, 148414.	1.0	15
21	mRNA-based therapy in a rabbit model of variegate porphyria offers new insights into the pathogenesis of acute attacks. Molecular Therapy - Nucleic Acids, 2021, 25, 207-219.	5.1	7
22	Distal phalangeal erythema in an infant with biallelic <scp>PDSS1</scp> mutations: Expanding the phenotype of primary Coenzyme <scp>Q₁₀</scp> deficiency. JIMD Reports, 2021, 62, 3-5.	1.5	4
23	Collaborative model for diagnosis and treatment of very rare diseases: experience in Spain with thymidine kinase 2 deficiency. Orphanet Journal of Rare Diseases, 2021, 16, 407.	2.7	3
24	The Genetic Landscape of Mitochondrial Diseases in Spain: A Nationwide Call. Genes, 2021, 12, 1590.	2.4	8
25	Regulation of Mitochondrial Function by the Actin Cytoskeleton. Frontiers in Cell and Developmental Biology, 2021, 9, 795838.	3.7	28
26	A novel mutation in the mitochondrial MT-ND5 gene in a family with MELAS. The relevance of genetic analysis on targeted tissues. Mitochondrion, 2020, 50, 14-18.	3.4	5
27	Creation and implementation of a European registry for patients with McArdle disease and other muscle glycogenoses (EUROMAC registry). Orphanet Journal of Rare Diseases, 2020, 15, 187.	2.7	3
28	Deoxynucleoside therapy for respiratory involvement in adult patients with thymidine kinase 2-deficient myopathy. BMJ Open Respiratory Research, 2020, 7, e000774.	3.0	9
29	Novel NDUFA13 Mutations Associated with OXPHOS Deficiency and Leigh Syndrome: A Second Family Report. Genes, 2020, 11, 855.	2.4	8
30	Biallelic mutations in NDUFA8 cause complex I deficiency in two siblings with favorable clinical evolution. Molecular Genetics and Metabolism, 2020, 131, 349-357.	1.1	6
31	Uniparental isodisomy as a cause of mitochondrial complex I respiratory chain disorder due to a novel splicing NDUFS4 mutation. Molecular Genetics and Metabolism, 2020, 131, 341-348.	1.1	10
32	Multiple pathways coordinate assembly of human mitochondrial complex IV and stabilization of respiratory supercomplexes. EMBO Journal, 2020, 39, e103912.	7.8	54
33	Altered Expression Ratio of Actin-Binding Gelsolin Isoforms Is a Novel Hallmark of Mitochondrial OXPHOS Dysfunction. Cells, 2020, 9, 1922.	4.1	11
34	Exercise Training and Neurodegeneration in Mitochondrial Disorders: Insights From the Harlequin Mouse. Frontiers in Physiology, 2020, 11, 594223.	2.8	4
35	Preclinical Research in Glycogen Storage Diseases: A Comprehensive Review of Current Animal Models. International Journal of Molecular Sciences, 2020, 21, 9621.	4.1	13
36	Mitochondrial Dysfunction and Calcium Dysregulation in Leigh Syndrome Induced Pluripotent Stem Cell Derived Neurons. International Journal of Molecular Sciences, 2020, 21, 3191.	4.1	19

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37	Growth Differentiation Factor 15 is a potential biomarker of therapeutic response for TK2 deficient myopathy. Scientific Reports, 2020, 10, 10111.	3.3	20
38	Sex Differences and the Influence of an Active Lifestyle on Adiposity in Patients with McArdle Disease. International Journal of Environmental Research and Public Health, 2020, 17, 4334.	2.6	2
39	Clinical, pathological and genetic spectrum in 89 cases of mitochondrial progressive external ophthalmoplegia. Journal of Medical Genetics, 2020, 57, 643-646.	3.2	21
40	Can routine laboratory variables predict survival in COVID-19? An artificial neural network-based approach. Clinical Chemistry and Laboratory Medicine, 2020, 58, e299-e302.	2.3	8
41	Physical Exercise and Mitochondrial Disease: Insights From a Mouse Model. Frontiers in Neurology, 2019, 10, 790.	2.4	15
42	P.124Is the expression of muscle glycogen phosphorylase tissue-specific? New perspectives on McArdle disease. Neuromuscular Disorders, 2019, 29, S84.	0.6	1
43	Novel ATAD3A recessive mutation associated to fatal cerebellar hypoplasia with multiorgan involvement and mitochondrial structural abnormalities. Molecular Genetics and Metabolism, 2019, 128, 452-462.	1.1	23
44	Genes and Variants Underlying Human Congenital Lactic Acidosisâ€"From Genetics to Personalized Treatment. Journal of Clinical Medicine, 2019, 8, 1811.	2.4	13
45	A Novel Missense Variant Associated with A Splicing Defect in A Myopathic Form of PGK1 Deficiency in The Spanish Population. Genes, 2019, 10, 785.	2.4	11
46	Late-onset thymidine kinase 2 deficiency: a review of 18 cases. Orphanet Journal of Rare Diseases, 2019, 14, 100.	2.7	29
47	Collagen XIX Alpha 1 Improves Prognosis in Amyotrophic Lateral Sclerosis. , 2019, 10, 278.		18
48	Low survival rate and muscle fiber-dependent aging effects in the McArdle disease mouse model. Scientific Reports, 2019, 9, 5116.	3.3	11
49	Increased dNTP pools rescue mtDNA depletion in human POLGâ€deficient fibroblasts. FASEB Journal, 2019, 33, 7168-7179.	0.5	14
50	Absence of p.R50X Pygm read-through in McArdle disease cellular models. DMM Disease Models and Mechanisms, 2019, 13, .	2.4	4
51	MiopatÃas metabÃ³licas, mitocondriales y tÃ³xicas. Medicine, 2019, 12, 4497-4506.	0.0	1
52	Defects in the mitochondrial-tRNA modification enzymes MTO1 and GTPBP3 promote different metabolic reprogramming through a HIF-PPARÎ³-UCP2-AMPK axis. Scientific Reports, 2018, 8, 1163.	3.3	23
53	Muscle molecular adaptations to endurance exercise training are conditioned by glycogen availability: a proteomicsâ€based analysis in the McArdle mouse model. Journal of Physiology, 2018, 596, 1035-1061.	2.9	26
54	Health Benefits of an Innovative Exercise Program for Mitochondrial Disorders. Medicine and Science in Sports and Exercise, 2018, 50, 1142-1151.	0.4	16

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55	Retrospective natural history of thymidine kinase 2 deficiency. Journal of Medical Genetics, 2018, 55, 515-521.	3.2	73
56	Nonâ€osteogenic muscle hypertrophy in children with McArdle disease. Journal of Inherited Metabolic Disease, 2018, 41, 1037-1042.	3.6	2
57	A New Condition in McArdle Disease. Medicine and Science in Sports and Exercise, 2018, 50, 3-10.	0.4	9
58	Las enfermedades raras en las patologÃas neurometabÃ³licas. Arbor, 2018, 194, 461.	0.3	0
59	The Mitochondrial Isoform of FASTK Modulates Nonopsonic Phagocytosis of Bacteria by Macrophages via Regulation of Respiratory Complex I. Journal of Immunology, 2018, 201, 2977-2985.	0.8	6
60	Manifesting heterozygotes in McArdle disease: a myth or a realityâ€"role of statins. Journal of Inherited Metabolic Disease, 2018, 41, 1027-1035.	3.6	4
61	Missense mutations have unexpected consequences: The McArdle disease paradigm. Human Mutation, 2018, 39, 1338-1343.	2.5	13
62	Different mitochondrial genetic defects exhibit the same protein signature of metabolism in skeletal muscle of PEO and MELAS patients: A role for oxidative stress. Free Radical Biology and Medicine, 2018, 126, 235-248.	2.9	10
63	The homozygous R504C mutation in MTO1 gene is responsible for ONCE syndrome. Clinical Genetics, 2017, 91, 46-53.	2.0	8
64	Respiratory chain enzyme deficiency induces mitochondrial location of actin-binding gelsolin to modulate the oligomerization of VDAC complexes and cell survival. Human Molecular Genetics, 2017, 26, 2493-2506.	2.9	13
65	Low versus high carbohydrates in the diet of the worldâ€class athlete: insights from McArdle's disease. Journal of Physiology, 2017, 595, 2991-2992.	2.9	1
66	Understanding mitochondrial diseases. Clinical Nutrition, 2017, 36, 902.	5.0	0
67	Rhodamine-based sensor for real-time imaging of mitochondrial ATP in living fibroblasts. Biochimica Et Biophysica Acta - Bioenergetics, 2017, 1858, 999-1006.	1.0	17
68	211th ENMC International Workshop:. Neuromuscular Disorders, 2017, 27, 1143-1151.	0.6	6
69	Establishment of a human iPSC line (IISHDOi001-A) from a patient with McArdle disease. Stem Cell Research, 2017, 23, 188-192.	0.7	4
70	Myopathic mtDNA Depletion Syndrome Due to Mutation in TK2 Gene. Pediatric and Developmental Pathology, 2017, 20, 416-420.	1.0	7
71	Muscle fiber type proportion and size is not altered in mcardle disease. Muscle and Nerve, 2017, 55, 916-918.	2.2	4
72	Genotypic and phenotypic features of all Spanish patients with McArdle disease: a 2016 update. BMC Genomics, 2017, 18, 819.	2.8	53

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73	Functional Characterization of Three Concomitant MtDNA LHON Mutations Shows No Synergistic Effect on Mitochondrial Activity. PLoS ONE, 2016, 11, e0146816.	2.5	17
74	Muscle Signaling in Exercise Intolerance. Medicine and Science in Sports and Exercise, 2016, 48, 1448-1458.	0.4	13
75	Secondary coenzyme Q 10 deficiencies in oxidative phosphorylation (OXPHOS) and non-OXPHOS disorders. Mitochondrion, 2016, 30, 51-58.	3.4	70
76	A milder phenotype of megaconial congenital muscular dystrophy due to a novelCHKBmutation. Muscle and Nerve, 2016, 54, 806-808.	2.2	3
77	Genes and exercise intolerance: insights from McArdle disease. Physiological Genomics, 2016, 48, 93-100.	2.3	15
78	In vivo evidence of mitochondrial dysfunction and altered redox homeostasis in a genetic mouse model of propionic acidemia: Implications for the pathophysiology of this disorder. Free Radical Biology and Medicine, 2016, 96, 1-12.	2.9	42
79	COX7A2L Is a Mitochondrial Complex III Binding Protein that Stabilizes the III2+IV Supercomplex without Affecting Respirasome Formation. Cell Reports, 2016, 16, 2387-2398.	6.4	93
80	New ATP8A2 gene mutations associated with a novel syndrome: encephalopathy, intellectual disability, severe hypotonia, chorea and optic atrophy. Neurogenetics, 2016, 17, 259-263.	1.4	36
81	Role of FAST Kinase Domains 3 (FASTKD3) in Post-transcriptional Regulation of Mitochondrial Gene Expression. Journal of Biological Chemistry, 2016, 291, 25877-25887.	3.4	37
82	Assessment of resting energy expenditure in pediatric mitochondrial diseases with indirect calorimetry. Clinical Nutrition, 2016, 35, 1484-1489.	5.0	8
83	Exercise and Preexercise Nutrition as Treatment for McArdle Disease. Medicine and Science in Sports and Exercise, 2016, 48, 673-679.	0.4	20
84	Taking advantage of an old concept, â€œillegitimate transcriptionâ€, for a proposed novel method of genetic diagnosis of McArdle disease. Genetics in Medicine, 2016, 18, 1128-1135.	2.4	9
85	First missense mutation outside of SERAC1 lipase domain affecting intracellular cholesterol trafficking. Neurogenetics, 2016, 17, 51-56.	1.4	7
86	Congenital neurogenic muscular atrophy in megaconial myopathy due to a mutation in CHKB gene. Brain and Development, 2016, 38, 167-172.	1.1	16
87	Xanthine Oxidase Pathway and Muscle Damage. Insights from McArdle Disease. Current Pharmaceutical Design, 2016, 22, 2657-2663.	1.9	2
88	A novel RRM2B gene variant associated with Telbivudine-induced mitochondrial myopathy. Journal of the Neurological Sciences, 2015, 358, 481-483.	0.6	7
89	Intracellular expression of Tat alters mitochondrial functions in T cells: a potential mechanism to understand mitochondrial damage during HIV-1 replication. Retrovirology, 2015, 12, 78.	2.0	27
90	McArdle Disease: Update of Reported Mutations and Polymorphisms in thePYGMGene. Human Mutation, 2015, 36, 669-678.	2.5	66

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91	Minimal symptoms in McArdle disease: A real PYGM genotype effect?. Muscle and Nerve, 2015, 52, 1136-1137.	2.2	1
92	iTRAQ-based analysis of progerin expression reveals mitochondrial dysfunction, reactive oxygen species accumulation and altered proteostasis. Stem Cell Research and Therapy, 2015, 6, 119.	5.5	28
93	Next-generation sequencing to estimate the prevalence of a great unknown: McArdle disease. Genetics in Medicine, 2015, 17, 679-680.	2.4	13
94	Phenotype consequences of myophosphorylase dysfunction: insights from the McArdle mouse model. Journal of Physiology, 2015, 593, 2693-2706.	2.9	17
95	Quantitative analysis of proteins of metabolism by reverse phase protein microarrays identifies potential biomarkers of rare neuromuscular diseases. Journal of Translational Medicine, 2015, 13, 65.	4.4	22
96	Sodium valproate increases the brain isoform of glycogen phosphorylase: looking for a compensation mechanism in McArdle disease using a mouse primary skeletal-muscle culture in vitro. DMM Disease Models and Mechanisms, 2015, 8, 467-472.	2.4	19
97	Severe TK2 enzyme activity deficiency in patients with mild forms of myopathy. Neurology, 2015, 84, 2286-2288.	1.1	26
98	Mitochondrial Respiration Controls Lysosomal Function during Inflammatory T Cell Responses. Cell Metabolism, 2015, 22, 485-498.	16.2	239
99	Differential proteomic profiling unveils new molecular mechanisms associated with mitochondrial complex III deficiency. Journal of Proteomics, 2015, 113, 38-56.	2.4	21
100	When should a nephrologist suspect a mitochondrial disease?. Nefrologia, 2015, 35, 6-17.	0.4	5
101	Follow-up of folinic acid supplementation for patients with cerebral folate deficiency and Kearns-Sayre syndrome. Orphanet Journal of Rare Diseases, 2014, 9, 217.	2.7	39
102	Exome sequencing identifies a CHKB mutation in Spanish patient with Megaconial Congenital Muscular Dystrophy and mtDNA depletion. European Journal of Paediatric Neurology, 2014, 18, 796-800.	1.6	21
103	Bulk autophagy, but not mitophagy, is increased in cellular model of mitochondrial disease. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2014, 1842, 1059-1070.	3.8	29
104	McArdle disease does not affect skeletal muscle fibre type profiles in humans. Biology Open, 2014, 3, 1224-1227.	1.2	8
105	Nitric oxide compounds have different effects profiles on human articular chondrocyte metabolism. Arthritis Research and Therapy, 2013, 15, R115.	3.5	38
106	A new mutation in the gene encoding mitochondrial seryl-tRNA synthetase as a cause of HUPRA syndrome. BMC Nephrology, 2013, 14, 195.	1.8	31
107	Whole-Exome Sequencing Identifies a Variant of the MitochondrialMT-ND1Gene Associated with Epileptic Encephalopathy: West Syndrome Evolving to Lennox-Gastaut Syndrome. Human Mutation, 2013, 34, 1623-1627.	2.5	25
108	Resistance (Weight Lifting) Training in an Adolescent With McArdle Disease. Journal of Child Neurology, 2013, 28, 805-808.	1.4	23

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109	The â€˜McArdle paradoxâ€™: exercise is a good advice for the exercise intolerant. British Journal of Sports Medicine, 2013, 47, 728-729.	6.7	17
110	Cardiac Dysfunction in Mitochondrial Disease. Circulation Journal, 2013, 77, 2799-2806.	1.6	5
111	Mitochondrial Complex III Deficiency of Nuclear Origin:. , 2013, , 219-238.		0
112	Knock-in mice for the R50X mutation in the PYGM gene present with McArdle disease. Brain, 2012, 135, 2048-2057.	7.6	48
113	Natural disease course and genotypeâ€phenotype correlations in Complex I deficiency caused by nuclear gene defects: what we learned from 130 cases. Journal of Inherited Metabolic Disease, 2012, 35, 737-747.	3.6	112
114	Nueva mutaciÃ³n en C10Orf2 (PEO1) en paciente con oftalmoplejÃa crÃ³nica progresiva mitocondrial dominante (adCPEO), primer caso argentino. Neurologia Argentina, 2012, 4, 157-161.	0.3	0
115	Mitochondrial tRNA valine as a recurrent target for mutations involved in mitochondrial cardiomyopathies. Mitochondrion, 2012, 12, 357-362.	3.4	15
116	Mitochondrial Complex I Plays an Essential Role in Human Respirasome Assembly. Cell Metabolism, 2012, 15, 324-335.	16.2	234
117	Mitochondrial respiratory chain dysfunction: Implications in neurodegeneration. Free Radical Biology and Medicine, 2012, 53, 595-609.	2.9	132
118	Genotypic and phenotypic features of McArdle disease: insights from the Spanish national registry. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 322-328.	1.9	114
119	Polymorphisms influencing muscle phenotypes in North-African and Spanish populations. Annals of Human Biology, 2012, 39, 166-169.	1.0	6
120	A Transcriptomic Approach to Search for Novel Phenotypic Regulators in McArdle Disease. PLoS ONE, 2012, 7, e31718.	2.5	22
121	Are mitochondrial haplogroups associated with extreme longevity? A study on a Spanish cohort. Age, 2012, 34, 227-233.	3.0	22
122	Are mitochondrial haplogroups associated with elite athletic status? A study on a Spanish cohort. Mitochondrion, 2011, 11, 905-908.	3.4	20
123	Effect of nitric oxide on mitochondrial activity of human synovial cells. BMC Musculoskeletal Disorders, 2011, 12, 42.	1.9	50
124	Primary Adenosine Monophosphate (AMP) Deaminase Deficiency in a Hypotonic Infant. Journal of Child Neurology, 2011, 26, 734-737.	1.4	9
125	Excessive skeletal muscle recruitment during strenuous exercise in McArdle patients. European Journal of Applied Physiology, 2010, 110, 1047-1055.	2.5	17
126	Cellular pathophysiological consequences of BCS1L mutations in mitochondrial complex III enzyme deficiency. Human Mutation, 2010, 31, 930-941.	2.5	61

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127	Impact of the Mitochondrial Genetic Background in Complex III Deficiency. PLoS ONE, 2010, 5, e12801.	2.5	34
128	Expression of Glycogen Phosphorylase Isoforms in Cultured Muscle from Patients with McArdle's Disease Carrying the p.R771PfsX33 PYGM Mutation. PLoS ONE, 2010, 5, e13164.	2.5	12
129	Diabetes and Mitochondrial Encephalomyopathy. , 2010, 20, 105-108.		0
130	A MELAS/MERRF phenotype associated with the mitochondrial DNA 5521G>A mutation. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, 471-472.	1.9	13
131	World-class performance in lightweight rowing: is it genetically influenced? A comparison with cyclists, runners and non-athletes. British Journal of Sports Medicine, 2010, 44, 898-901.	6.7	71
132	Marked mitochondrial DNA depletion associated with a novel SUCLG1 gene mutation resulting in lethal neonatal acidosis, multi-organ failure, and interrupted aortic arch. Mitochondrion, 2010, 10, 362-368.	3.4	29
133	Kearns-Sayre syndrome: Cerebral folate deficiency, MRI findings and new cerebrospinal fluid biochemical features. Mitochondrion, 2010, 10, 429-432.	3.4	53
134	Mitochondrial bioenergetics and dynamics interplay in complex I-deficient fibroblasts. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2010, 1802, 443-453.	3.8	66
135	AÎ² accumulation in choroid plexus is associated with mitochondrial-induced apoptosis. Neurobiology of Aging, 2010, 31, 1569-1581.	3.1	63
136	Genotype Distributions in Top-level Soccer Players: A Role for<i>ACE</i>?. International Journal of Sports Medicine, 2009, 30, 387-392.	1.7	43
137	Coenzyme Q10 deficiency associated with a mitochondrial DNA depletion syndrome: A case report. Clinical Biochemistry, 2009, 42, 742-745.	1.9	25
138	Infantile mitochondrial encephalomyopathy with unusual phenotype caused by a novel BCS1L mutation in an isolated complex III-deficient patient. Neuromuscular Disorders, 2009, 19, 143-146.	0.6	44
139	Pathogenic mutations in the 5â€² untranslated region of BCS1L mRNA in mitochondrial complex III deficiency. Mitochondrion, 2009, 9, 299-305.	3.4	29
140	Mitochondrial Disorders Due to Nuclear OXPHOS Gene Defects. Advances in Experimental Medicine and Biology, 2009, 652, 85-116.	1.6	9
141	Mitochondrial Encephalomyopathies in Children. Part I: Conventional MR Imaging Findings. Current Medical Imaging, 2009, 5, 85-99.	0.8	1
142	Mitochondrial Encephalomyopathies in Children. Part II: Advanced MR Tools and the Importance for its Early Recognition in the Acute Clinical Setting. Current Medical Imaging, 2009, 5, 100-109.	0.8	0
143	C34T mutation of the AMPD1 gene in an elite white runner. BMJ Case Reports, 2009, 2009, bcr0720080535-bcr0720080535.	0.5	8
144	Expression of the muscle glycogen phosphorylase gene in patients with McArdle disease: the role of nonsense-mediated mRNA decay. Human Mutation, 2008, 29, 277-283.	2.5	38

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145	McArdle disease: Another systemic low-inflammation disorder?. Neuroscience Letters, 2008, 431, 106-111.	2.1	9
146	McArdle disease: what do neurologists need to know?. Nature Clinical Practice Neurology, 2008, 4, 568-577.	2.5	195
147	OPA1 mutations induce mitochondrial DNA instability and optic atrophy 'plus' phenotypes. Brain, 2008, 131, 338-351.	7.6	454
148	Novel mutations in patients with McArdle disease by analysis of skeletal muscle mRNA. Journal of Medical Genetics, 2008, 46, 198-202.	3.2	20
149	Mitochondrial DNA background modulates the assembly kinetics of OXPHOS complexes in a cellular model of mitochondrial disease. Human Molecular Genetics, 2008, 17, 4001-4011.	2.9	140
150	AMPD1 Genotypes and Exercise Capacity in McArdle Patients. International Journal of Sports Medicine, 2008, 29, 331-335.	1.7	11
151	The I allele of the ACE gene is associated with improved exercise capacity in women with McArdle disease. British Journal of Sports Medicine, 2007, 42, 134-140.	6.7	19
152	Exercise Capacity in a Child With McArdle Disease. Journal of Child Neurology, 2007, 22, 880-882.	1.4	15
153	Favorable Responses to Acute and Chronic Exercise in McArdle Patients. Clinical Journal of Sport Medicine, 2007, 17, 297-303.	1.8	85
154	The 577X allele of the ACTN3 gene is associated with improved exercise capacity in women with McArdleâ€™s disease. Neuromuscular Disorders, 2007, 17, 603-610.	0.6	32
155	Mild ocular myopathy associated with a novel mutation in mitochondrial twinkle helicase. Neuromuscular Disorders, 2007, 17, 677-680.	0.6	14
156	Genotype modulators of clinical severity in McArdle disease. Neuroscience Letters, 2007, 422, 217-222.	2.1	40
157	X-linkedNDUFA1gene mutations associated with mitochondrial encephalomyopathy. Annals of Neurology, 2007, 61, 73-83.	5.3	118
158	A proposed molecular diagnostic flowchart for myophosphorylase deficiency (McArdle disease) in blood samples from Spanish patients. Human Mutation, 2007, 28, 203-204.	2.5	54
159	C34T mutation of the AMPD1 gene in an elite white runner. British Journal of Sports Medicine, 2006, 40, e7-e7.	6.7	15
160	Mitochondrial activity is modulated by TNFÎ± and IL-1Î² in normal human chondrocyte cells. Osteoarthritis and Cartilage, 2006, 14, 1011-1022.	1.3	121
161	Does the C34T Mutation in AMPD1 Alter Exercise Capacity in the Elderly?. International Journal of Sports Medicine, 2006, 27, 429-435.	1.7	6
162	Mobilisation of mesenchymal cells into blood in response to skeletal muscle injury. British Journal of Sports Medicine, 2006, 40, 719-722.	6.7	53

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163	Association of Novel POLGMutations and Multiple Mitochondrial DNA Deletions With Variable Clinical Phenotypes in a Spanish Population. Archives of Neurology, 2006, 63, 107.	4.5	57
164	Can patients with McArdle's disease run? * Commentary. British Journal of Sports Medicine, 2006, 41, 53-54.	6.7	19
165	Novel Mutation in the PYGM Gene Resulting in McArdle Disease. Archives of Neurology, 2006, 63, 1782.	4.5	9
166	Exercise capacity in a 78 year old patient with McArdle's disease: it is never too late to start exercising * Commentary. British Journal of Sports Medicine, 2006, 40, 725-726.	6.7	8
167	Chronic primary intestinal pseudo-obstruction from visceral myopathy. Revista Espanola De Enfermedades Digestivas, 2006, 98, 292-302.	0.3	11
168	Renal pathology in children with mitochondrial diseases. Pediatric Nephrology, 2005, 20, 1299-1305.	1.7	105
169	Leigh Syndrome Associated With Mitochondrial Complex I Deficiency Due to a Novel Mutation in the NDUFS1 Gene. Archives of Neurology, 2005, 62, 659.	4.5	71
170	Frequency of the C34T mutation of the AMPD1 gene in world-class endurance athletes: does this mutation impair performance?. Journal of Applied Physiology, 2005, 98, 2108-2112.	2.5	76
171	Comparative analysis of the pathogenic mechanisms associated with the G8363A and A8296G mutations in the mitochondrial tRNALys gene. Biochemical Journal, 2005, 387, 773-778.	3.7	27
172	Increased muscle nucleoside levels associated with a novel frameshift mutation in the thymidine phosphorylase gene in a Spanish patient with MNGIE. Neuromuscular Disorders, 2005, 15, 775-778.	0.6	18
173	Effect of nitric oxide on mitochondrial respiratory activity of human articular chondrocytes. Annals of the Rheumatic Diseases, 2004, 64, 388-395.	0.9	122
174	Molecular Analysis of Myophosphorylase Deficiency in Dutch Patients with McArdle's Disease. Annals of Human Genetics, 2004, 68, 17-22.	0.8	26
175	Apolipoprotein E polymorphism and carotid atherosclerosis in patients with coronary disease. International Journal of Cardiology, 2004, 94, 209-212.	1.7	17
176	Defective hepatic mitochondrial respiratory chain in patients with nonalcoholic steatohepatitis. Hepatology, 2003, 38, 999-1007.	7.3	585
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