Joaquin Arenas

List of Publications by Year
in descending order

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186 papers	7,561 citations	61687 45 h-index	75989 78 g-index
192 all docs	192 docs citations	192 times ranked	9393 citing authors

#	Article	IF	Citations
1	Defective hepatic mitochondrial respiratory chain in patients with nonalcoholic steatohepatitis. Hepatology, 2003, 38, 999-1007.	3.6	585
2	OPA1 mutations induce mitochondrial DNA instability and optic atrophy 'plus' phenotypes. Brain, 2008, 131, 338-351.	3.7	454
3	Defective hepatic mitochondrial respiratory chain in patients with nonalcoholic steatohepatitis. Hepatology, 2003, 38, 999-1007.	3.6	358
4	Mitochondrial Complex I Plays an Essential Role in Human Respirasome Assembly. Cell Metabolism, 2012, 15, 324-335.	7.2	234
5	Mitochondrial respiratory activity is altered in osteoarthritic human articular chondrocytes. Arthritis and Rheumatism, 2003, 48, 700-708.	6.7	195
6	McArdle disease: what do neurologists need to know?. Nature Clinical Practice Neurology, 2008, 4, 568-577.	2.7	195
7	Sirolimus Does Not Exhibit Nephrotoxicity Compared to Cyclosporine in Renal Transplant Recipients. American Journal of Transplantation, 2002, 2, 436-442.	2.6	178
8	Complex I Defect in muscle from patients with Huntington's disease. Annals of Neurology, 1998, 43, 397-400.	2.8	154
9	Specific correlation between the wobble modification deficiency in mutant tRNAs and the clinical features of a human mitochondrial disease. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 7127-7132.	3.3	147
10	Mitochondrial DNA background modulates the assembly kinetics of OXPHOS complexes in a cellular model of mitochondrial disease. Human Molecular Genetics, 2008, 17, 4001-4011.	1.4	140
11	Mitochondrial respiratory chain dysfunction: Implications in neurodegeneration. Free Radical Biology and Medicine, 2012, 53, 595-609.	1.3	132
12	Effect of nitric oxide on mitochondrial respiratory activity of human articular chondrocytes. Annals of the Rheumatic Diseases, 2004, 64, 388-395.	0.5	122
13	Mitochondrial activity is modulated by TNFÎ± and IL-1Î² in normal human chondrocyte cells. Osteoarthritis and Cartilage, 2006, 14, 1011-1022.	0.6	121
14	X-linkedNDUFA1gene mutations associated with mitochondrial encephalomyopathy. Annals of Neurology, 2007, 61, 73-83.	2.8	118
15	Genotypic and phenotypic features of McArdle disease: insights from the Spanish national registry. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 322-328.	0.9	114
16	Renal pathology in children with mitochondrial diseases. Pediatric Nephrology, 2005, 20, 1299-1305.	0.9	105
17	Mitochondrial DNA haplogroups: Role in the prevalence and severity of knee osteoarthritis. Arthritis and Rheumatism, 2008, 58, 2387-2396.	6.7	96
18	COX7A2L Is a Mitochondrial Complex III Binding Protein that Stabilizes the III2+IV Supercomplex without Affecting Respirasome Formation. Cell Reports, 2016, 16, 2387-2398.	2.9	93

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19	Plasma carnitine insufficiency and effectiveness ofL-carnitine therapy in patients with mitochondril myopathy. Muscle and Nerve, 1993, 16, 150-153.	1.0	91
20	Exercise training in mitochondrial myopathy: A randomized controlled trial. Muscle and Nerve, 2005, 32, 342-350.	1.0	87
21	Molecular heterogeneity of myophosphorylase deficiency (Mcardle's disease): A genotype-phenotype correlation study. Annals of Neurology, 2001, 50, 574-581.	2.8	86
22	Favorable Responses to Acute and Chronic Exercise in McArdle Patients. Clinical Journal of Sport Medicine, 2007, 17, 297-303.	0.9	85
23	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.	1.2	76
24	Nephrotic Proteinuria Without Hypoalbuminemia: Clinical Characteristics and Response to Angiotensin-Converting Enzyme Inhibition. American Journal of Kidney Diseases, 1991, 17, 330-338.	2.1	75
25	Serum levels of beta-carotene, alpha-carotene and vitamin A in patients with Alzheimer's disease. European Journal of Neurology, 1999, 6, 495-497.	1.7	71
26	Leigh Syndrome Associated With Mitochondrial Complex I Deficiency Due to a Novel Mutation in the NDUFS1 Gene. Archives of Neurology, 2005, 62, 659.	4.9	71
27	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.	3.1	71
28	Secondary coenzyme Q 10 deficiencies in oxidative phosphorylation (OXPHOS) and non-OXPHOS disorders. Mitochondrion, 2016, 30, 51-58.	1.6	70
29	McArdle Disease: Update of Reported Mutations and Polymorphisms in thePYGMGene. Human Mutation, 2015, 36, 669-678.	1.1	66
30	Cellular pathophysiological consequences of BCS1L mutations in mitochondrial complex III enzyme deficiency. Human Mutation, 2010, 31, 930-941.	1.1	61
31	Oxidative Stress in Skin Fibroblasts Cultures of Patients with Huntingtonâ€™s Disease. Neurochemical Research, 2006, 31, 1103-1109.	1.6	57
32	Association of Novel POLGMutations and Multiple Mitochondrial DNA Deletions With Variable Clinical Phenotypes in a Spanish Population. Archives of Neurology, 2006, 63, 107.	4.9	57
33	Neurotransmitter amino acids in cerebrospinal fluid of patients with Parkinson's disease. Journal of the Neurological Sciences, 1996, 141, 39-44.	0.3	56
34	Amylase Levels in Pleural Effusions. Chest, 2002, 121, 470-474.	0.4	56
35	Two homozygous mutations (R193W and 794/795 delAA) in the myophosphorylase gene in a patient with McArdle's disease. Human Mutation, 2000, 15, 294-294.	1.1	55
36	A proposed molecular diagnostic flowchart for myophosphorylase deficiency (McArdle disease) in blood samples from Spanish patients. Human Mutation, 2007, 28, 203-204.	1.1	54

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37	Multiple pathways coordinate assembly of human mitochondrial complex IV and stabilization of respiratory supercomplexes. EMBO Journal, 2020, 39, e103912.	3.5	54
38	Mobilisation of mesenchymal cells into blood in response to skeletal muscle injury. British Journal of Sports Medicine, 2006, 40, 719-722.	3.1	53
39	Genotypic and phenotypic features of all Spanish patients with McArdle disease: a 2016 update. BMC Genomics, 2017, 18, 819.	1.2	53
40	Mitochondrial DNA deletion in a patient with mitochondrial myopathy, lactic acidosis, and stroke-like episodes (MELAS) and Fanconi's syndrome. Pediatric Neurology, 1995, 13, 69-72.	1.0	51
41	Early onset multisystem mitochondrial disorder caused by a nonsense mutation in the mitochondrial DNACytochrome C oxidase IIgene. Annals of Neurology, 2001, 50, 409-413.	2.8	51
42	Thyroid hormone regulates oxidative phosphorylation in the cerebral cortex and striatum of neonatal rats. Journal of Neurochemistry, 2001, 78, 1054-1063.	2.1	50
43	Effect of nitric oxide on mitochondrial activity of human synovial cells. BMC Musculoskeletal Disorders, 2011, 12, 42.	0.8	50
44	Syndrome of encephalopathy, petechiae, and ethylmalonic aciduria. Pediatric Neurology, 1997, 17, 165-170.	1.0	48
45	Knock-in mice for the R50X mutation in the PYGM gene present with McArdle disease. Brain, 2012, 135, 2048-2057.	3.7	48
46	Mitochondrial DNA Haplogroups Do Not Play a Role in the Variable Phenotypic Presentation of the A3243G Mutation. American Journal of Human Genetics, 2003, 72, 1005-1012.	2.6	47
47	Effects ofl-carnitine on the pyruvate dehydrogenase complex and carnitine palmitoyl transferase activities in muscle of endurance athletes. FEBS Letters, 1994, 341, 91-93.	1.3	44
48	Lipomatosis, proximal myopathy, and the mitochondrial 8344 mutation. A lipid storage myopathy?. , 2000, 23, 538-542.		44
49	Prevalence and progression of mitochondrial diseases: A study of 50 patients. Muscle and Nerve, 2003, 28, 690-695.	1.0	44
50	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.	0.3	44
51	Strenuous endurance exercise improves life expectancy: it's in our genes. British Journal of Sports Medicine, 2011, 45, 159-161.	3.1	43
52	Respiratory chain enzymes in muscle of endurance athletes: Effect of L-carnitine. Biochemical and Biophysical Research Communications, 1992, 188, 102-107.	1.0	40
53	Genotype modulators of clinical severity in McArdle disease. Neuroscience Letters, 2007, 422, 217-222.	1.0	40
54	Muscle carnitine deficiency and lipid storage myopathy in patients with mitochondrial myopathy. Muscle and Nerve, 1993, 16, 778-781.	1.0	39

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55	Follow-up of folinic acid supplementation for patients with cerebral folate deficiency and Kearns-Sayre syndrome. Orphanet Journal of Rare Diseases, 2014, 9, 217.	1.2	39
56	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.	1.1	38
57	Nitric oxide compounds have different effects profiles on human articular chondrocyte metabolism. Arthritis Research and Therapy, 2013, 15, R115.	1.6	38
58	Oxidative stress in skin fibroblasts cultures from patients with Parkinson's disease. BMC Neurology, 2010, 10, 95.	0.8	37
59	Sporadic MERRF/MELAS overlap syndrome associated with the 3243 tRNALeu(UUR) mutation of mitochondrial DNA. , 1996, 19, 187-190.		36
60	New ATP8A2 gene mutations associated with a novel syndrome: encephalopathy, intellectual disability, severe hypotonia, chorea and optic atrophy. Neurogenetics, 2016, 17, 259-263.	0.7	36
61	Impact of the Mitochondrial Genetic Background in Complex III Deficiency. PLoS ONE, 2010, 5, e12801.	1.1	34
62	Free radicals-mediated damage in transmitochondrial cells harboring the T14487C mutation in the ND6 gene of mtDNA. FEBS Letters, 2005, 579, 6909-6913.	1.3	32
63	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.	0.3	32
64	Diffuse Fatty Liver in Familial Heterozygous Hypobetalipoproteinemia. Journal of Clinical Gastroenterology, 1997, 25, 379-382.	1.1	32
65	A new mutation in the gene encoding mitochondrial seryl-tRNA synthetase as a cause of HUPRA syndrome. BMC Nephrology, 2013, 14, 195.	0.8	31
66	About the â€œPathologicalâ€•Role of the mtDNA T3308C Mutationâ€¦. American Journal of Human Genetics, 1999, 65, 1457-1459.	2.6	30
67	Pathogenic mutations in the 5â€² untranslated region of BCS1L mRNA in mitochondrial complex III deficiency. Mitochondrion, 2009, 9, 299-305.	1.6	29
68	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.	1.6	29
69	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.	1.8	29
70	Late-onset thymidine kinase 2 deficiency: a review of 18 cases. Orphanet Journal of Rare Diseases, 2019, 14, 100.	1.2	29
71	Regulation of Mitochondrial Function by the Actin Cytoskeleton. Frontiers in Cell and Developmental Biology, 2021, 9, 795838.	1.8	28
72	Identification of novel mutations in Spanish patients with muscle carnitine palmitoyltransferase II deficiency. Human Mutation, 2000, 15, 579-580.	1.1	27

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73	Comparative analysis of the pathogenic mechanisms associated with the G8363A and A8296G mutations in the mitochondrial tRNALys gene. Biochemical Journal, 2005, 387, 773-778.	1.7	27
74	Molecular Analysis of Myophosphorylase Deficiency in Dutch Patients with McArdle's Disease. Annals of Human Genetics, 2004, 68, 17-22.	0.3	26
75	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.	1.3	26
76	Double trouble (McArdle's disease and myasthenia gravis): How can exercise help?. Muscle and Nerve, 2007, 35, 125-128.	1.0	25
77	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.	1.1	25
78	Congenital Hydranencephalic-Hydrocephalic Syndrome With Proliferative Vasculopathy: A Possible Relation With Mitochondrial Dysfunction. Journal of Child Neurology, 2001, 16, 858-862.	0.7	24
79	Physical exercise and epicardial adipose tissue: A systematic review and metaâ€analysis of randomized controlled trials. Obesity Reviews, 2021, 22, e13103.	3.1	24
80	Association of genetically proven deficiencies of myophosphorylase and AMP deaminase: a second case of â€˜double troubleâ€™. Neuromuscular Disorders, 1997, 7, 387-389.	0.3	23
81	Novel ATAD3A recessive mutation associated to fatal cerebellar hypoplasia with multiorgan involvement and mitochondrial structural abnormalities. Molecular Genetics and Metabolism, 2019, 128, 452-462.	0.5	23
82	Congenital Hydranencephalic-Hydrocephalic Syndrome Associated With Mitochondrial Dysfunction. Journal of Child Neurology, 1999, 14, 131-135.	0.7	22
83	Cerebrospinal Fluid Nitrate Levels in Patients with Multiple Sclerosis. European Neurology, 1999, 41, 44-47.	0.6	22
84	Molecular analysis in spanish patients with muscle carnitine palmitoyltransferase deficiency. , 1999, 22, 941-943.		22
85	Human mitochondrial pyrophosphatase: cDNA cloning and analysis of the gene in patients with mtDNA depletion syndromes. Genomics, 2006, 87, 410-416.	1.3	22
86	A Transcriptomic Approach to Search for Novel Phenotypic Regulators in McArdle Disease. PLoS ONE, 2012, 7, e31718.	1.1	22
87	Are mitochondrial haplogroups associated with extreme longevity? A study on a Spanish cohort. Age, 2012, 34, 227-233.	3.0	22
88	Molecular analysis of Spanish patients with AMP deaminase deficiency. Muscle and Nerve, 2000, 23, 1175-1178.	1.0	21
89	Are elite endurance athletes genetically predisposed to lower disease risk?. Physiological Genomics, 2010, 41, 82-90.	1.0	21
90	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.	0.7	21

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91	Differential proteomic profiling unveils new molecular mechanisms associated with mitochondrial complex III deficiency. Journal of Proteomics, 2015, 113, 38-56.	1.2	21
92	Clinical, pathological and genetic spectrum in 89 cases of mitochondrial progressive external ophthalmoplegia. Journal of Medical Genetics, 2020, 57, 643-646.	1.5	21
93	Reactive oxygen species mediate the down-regulation of mitochondrial transcripts and proteins by tumour necrosis factor-alpha in L929 cells. Biochemical Journal, 2003, 370, 609-619.	1.7	20
94	Novel mutations in patients with McArdle disease by analysis of skeletal muscle mRNA. Journal of Medical Genetics, 2008, 46, 198-202.	1.5	20
95	Are mitochondrial haplogroups associated with elite athletic status? A study on a Spanish cohort. Mitochondrion, 2011, 11, 905-908.	1.6	20
96	Exercise and Preexercise Nutrition as Treatment for McArdle Disease. Medicine and Science in Sports and Exercise, 2016, 48, 673-679.	0.2	20
97	Can patients with McArdle's disease run? * Commentary. British Journal of Sports Medicine, 2006, 41, 53-54.	3.1	19
98	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.	3.1	19
99	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.	1.2	19
100	Mitochondrial myopathy, cardiomyopathy and psychiatric illness in a Spanish family harbouring the mtDNA 3303C > T mutation. Journal of Inherited Metabolic Disease, 2001, 24, 685-687.	1.7	18
101	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.	0.3	18
102	Mitochondrial gene expression and respiratory enzyme activities in cardiac diseases. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 1998, 1406, 85-90.	1.8	17
103	Apolipoprotein E polymorphism and carotid atherosclerosis in patients with coronary disease. International Journal of Cardiology, 2004, 94, 209-212.	0.8	17
104	One-Year Follow-Up in a Child With McArdle Disease: Exercise is Medicine. Pediatric Neurology, 2008, 38, 133-136.	1.0	17
105	Excessive skeletal muscle recruitment during strenuous exercise in McArdle patients. European Journal of Applied Physiology, 2010, 110, 1047-1055.	1.2	17
106	Phenotype consequences of myophosphorylase dysfunction: insights from the McArdle mouse model. Journal of Physiology, 2015, 593, 2693-2706.	1.3	17
107	Plasma levels of nitrates in patients with Parkinson's disease. Journal of the Neurological Sciences, 1994, 127, 87-89.	0.3	16
108	Congenital neurogenic muscular atrophy in megaconial myopathy due to a mutation in CHKB gene. Brain and Development, 2016, 38, 167-172.	0.6	16

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109	Health Benefits of an Innovative Exercise Program for Mitochondrial Disorders. Medicine and Science in Sports and Exercise, 2018, 50, 1142-1151.	0.2	16
110	Resolution of a mispaired secondary structure intermediate could account for a novel micro-insertion/deletion (387 insA/del 8 bp) in the PYGM gene causing McArdle's disease. Clinical Genetics, 2001, 59, 48-51.	1.0	15
111	The A8296G mtDNA mutation associated with several mitochondrial diseases does not cause mitochondrial dysfunction in cybrid cell lines. Human Mutation, 2002, 19, 234-239.	1.1	15
112	Expression of glucose transporter-2, glucokinase and mitochondrial glycerolphosphate dehydrogenase in pancreatic islets during rat ontogenesis. FEBS Journal, 2002, 269, 119-127.	0.2	15
113	Exercise Capacity in a Child With McArdle Disease. Journal of Child Neurology, 2007, 22, 880-882.	0.7	15
114	Mitochondrial tRNA valine as a recurrent target for mutations involved in mitochondrial cardiomyopathies. Mitochondrion, 2012, 12, 357-362.	1.6	15
115	Genes and exercise intolerance: insights from McArdle disease. Physiological Genomics, 2016, 48, 93-100.	1.0	15
116	Physical Exercise and Mitochondrial Disease: Insights From a Mouse Model. Frontiers in Neurology, 2019, 10, 790.	1.1	15
117	SILAC-based complexome profiling dissects the structural organization of the human respiratory supercomplexes in SCAFIKO cells. Biochimica Et Biophysica Acta - Bioenergetics, 2021, 1862, 148414.	0.5	15
118	A missense mutation W797R in the myophosphorylase gene in a Spanish patient with McArdle's disease. Muscle and Nerve, 2000, 23, 129-131.	1.0	14
119	Increased mitochondrial respiratory chain enzyme activities correlate with minor extent of liver damage in mice suffering from erythropoietic protoporphyria. Experimental Dermatology, 2005, 14, 26-33.	1.4	14
120	Mild ocular myopathy associated with a novel mutation in mitochondrial twinkle helicase. Neuromuscular Disorders, 2007, 17, 677-680.	0.3	14
121	Next-generation sequencing to estimate the prevalence of a great unknown: McArdle disease. Genetics in Medicine, 2015, 17, 679-680.	1.1	13
122	Muscle Signaling in Exercise Intolerance. Medicine and Science in Sports and Exercise, 2016, 48, 1448-1458.	0.2	13
123	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.	1.4	13
124	Missense mutations have unexpected consequences: The McArdle disease paradigm. Human Mutation, 2018, 39, 1338-1343.	1.1	13
125	Preclinical Research in Glycogen Storage Diseases: A Comprehensive Review of Current Animal Models. International Journal of Molecular Sciences, 2020, 21, 9621.	1.8	13
126	Cyclosporine Nephrotoxicity and Rejection Crisis: Diagnosis by Urinary Enzyme Excretion. Nephron, 1996, 72, 402-406.	0.9	12

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127	Cosegregation of the mitochondrial DNA A1555G and G4309A mutations results in deafness and mitochondrial myopathy. Muscle and Nerve, 2002, 25, 185-188.	1.0	12
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.	1.1	12
129	Cerebrospinal fluid carnitine levels in patients with Parkinson's disease. Journal of the Neurological Sciences, 1997, 145, 183-185.	0.3	11
130	Low survival rate and muscle fiber-dependent aging effects in the McArdle disease mouse model. Scientific Reports, 2019, 9, 5116.	1.6	11
131	Altered Expression Ratio of Actin-Binding Gelsolin Isoforms Is a Novel Hallmark of Mitochondrial OXPHOS Dysfunction. Cells, 2020, 9, 1922.	1.8	11
132	Abnormal carnitine distribution in the muscles of patients with idiopathic inflammatory myopathy. Arthritis and Rheumatism, 1996, 39, 1869-1874.	6.7	10
133	Two novel mutations in the muscle glycogen phosphorylase gene in McArdle's disease. Muscle and Nerve, 2003, 28, 380-382.	1.0	9
134	Novel Mutation in the PYGM Gene Resulting in McArdle Disease. Archives of Neurology, 2006, 63, 1782.	4.9	9
135	McArdle disease: Another systemic low-inflammation disorder?. Neuroscience Letters, 2008, 431, 106-111.	1.0	9
136	The second wind phenomenon in very young McArdleâ€™s patients. Neuromuscular Disorders, 2009, 19, 403-405.	0.3	9
137	Mitochondrial Disorders Due to Nuclear OXPHOS Gene Defects. Advances in Experimental Medicine and Biology, 2009, 652, 85-116.	0.8	9
138	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.	1.1	9
139	A New Condition in McArdle Disease. Medicine and Science in Sports and Exercise, 2018, 50, 3-10.	0.2	9
140	Muscle carnitine deficiency associated with zidovudine-induced mitochondrial myopathy. Annals of Neurology, 1994, 36, 680-680.	2.8	8
141	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.	3.1	8
142	McArdle disease does not affect skeletal muscle fibre type profiles in humans. Biology Open, 2014, 3, 1224-1227.	0.6	8
143	Assessment of resting energy expenditure in pediatric mitochondrial diseases with indirect calorimetry. Clinical Nutrition, 2016, 35, 1484-1489.	2.3	8
144	Plasma Gelsolin Reinforces the Diagnostic Value of FGF-21 and GDF-15 for Mitochondrial Disorders. International Journal of Molecular Sciences, 2021, 22, 6396.	1.8	8

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145	C34T mutation of the AMPD1 gene in an elite white runner. BMJ Case Reports, 2009, 2009, bcr0720080535-bcr0720080535.	0.2	8
146	Reduced carnitine palmitoyl transferase activity and altered acyl-trafficking in red blood cells from hemodialysis patients. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 1996, 1315, 37-39.	1.8	7
147	The association of Acetyl-l-Carnitine with glucose and lipid metabolism in human muscle in vivo: The effect of hyperinsulinemia. Metabolism: Clinical and Experimental, 1997, 46, 1454-1457.	1.5	7
148	The V368i mutation in Twinkle does not segregate with adPEO. Annals of Neurology, 2003, 53, 278-278.	2.8	7
149	Unusual clinical findings and Complex III deficiency in a family with myotonic dystrophy. Journal of the Neurological Sciences, 2003, 208, 87-91.	0.3	7
150	A novel RRM2B gene variant associated with Telbivudine-induced mitochondrial myopathy. Journal of the Neurological Sciences, 2015, 358, 481-483.	0.3	7
151	First missense mutation outside of SERAC1 lipase domain affecting intracellular cholesterol trafficking. Neurogenetics, 2016, 17, 51-56.	0.7	7
152	Myopathic mtDNA Depletion Syndrome Due to Mutation in TK2 Gene. Pediatric and Developmental Pathology, 2017, 20, 416-420.	0.5	7
153	Elevated glutamate and decreased glutamine levels in the cerebrospinal fluid of patients with MELAS syndrome. Journal of Neurology, 2022, 269, 3238-3248.	1.8	7
154	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.	0.2	7
155	What isn't taught in medical schools: the William Wordsworth lesson. Nature Clinical Practice Cardiovascular Medicine, 2008, 5, 372-374.	3.3	6
156	Does the K153R variant of the myostatin gene influence the clinical presentation of women with McArdle disease?. Neuromuscular Disorders, 2009, 19, 220-222.	0.3	6
157	Clinical and cellular consequences of the mutation m.12300G>A in the mitochondrial tRNALeu(CUN) gene. Mitochondrion, 2012, 12, 288-293.	1.6	6
158	Exercise Benefits in Pulmonary Hypertension. Journal of the American College of Cardiology, 2019, 73, 2906-2907.	1.2	5
159	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.	1.0	5
160	Macro creatine kinase type 2 in a patient with prostatic carcinoma. Clinica Chimica Acta, 1991, 200, 53-56.	0.5	4
161	Leber's Congenital Amaurosis Associated With Mitochondrial Dysfunction. Journal of Child Neurology, 1996, 11, 108-111.	0.7	4
162	Muscle fiber type proportion and size is not altered in mcardle disease. Muscle and Nerve, 2017, 55, 916-918.	1.0	4

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163	Manifesting heterozygotes in McArdle disease: a myth or a realityâ€"role of statins. Journal of Inherited Metabolic Disease, 2018, 41, 1027-1035.	1.7	4
164	Absence of p.R50X Pygm read-through in McArdle disease cellular models. DMM Disease Models and Mechanisms, 2019, 13, .	1.2	4
165	Exercise Training and Neurodegeneration in Mitochondrial Disorders: Insights From the Harlequin Mouse. Frontiers in Physiology, 2020, 11, 594223.	1.3	4
166	Preclinical Research in McArdle Disease: A Review of Research Models and Therapeutic Strategies. Genes, 2022, 13, 74.	1.0	4
167	Serum levels of nitrates in focal cerebral ischemia. European Journal of Neurology, 1995, 2, 225-227.	1.7	3
168	A milder phenotype of megaconial congenital muscular dystrophy due to a novelCHKBmutation. Muscle and Nerve, 2016, 54, 806-808.	1.0	3
169	Letters to the editor. Muscle and Nerve, 1992, 15, 1055-1059.	1.0	2
170	Letters to the editor. Muscle and Nerve, 1994, 17, 1225-1238.	1.0	2
171	Multiple deletions of mitochondrial DNA in muscle from a patient with benign progressive external ophthalmoplegia. Journal of Inherited Metabolic Disease, 1996, 19, 366-367.	1.7	2
172	Slow segregation and rapid shift to homoplasmy coexist in a family with the T8993>G mutation. Journal of Inherited Metabolic Disease, 1999, 22, 939-940.	1.7	2
173	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.	1.2	2
174	Xanthine Oxidase Pathway and Muscle Damage. Insights from McArdle Disease. Current Pharmaceutical Design, 2016, 22, 2657-2663.	0.9	2
175	Metrics of Progression and Prognosis in Untreated Adults With Thymidine Kinase 2 Deficiency: An Observational Study. Neuromuscular Disorders, 2022, , .	0.3	2
176	Mitochondrial Encephalomyopathies in Children. Part I: Conventional MR Imaging Findings. Current Medical Imaging, 2009, 5, 85-99.	0.4	1
177	Minimal symptoms in McArdle disease: A real PYGM genotype effect?. Muscle and Nerve, 2015, 52, 1136-1137.	1.0	1
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