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	Defective hepatic mitochondrial respiratory chain in patients with nonalcoholic steatohepatitis. Hepatology, 2003, 38, 999-1007.	7.3	585
2	OPA1 mutations induce mitochondrial DNA instability and optic atrophy 'plus' phenotypes. Brain, 2008, 131, 338-351.	7.6	454
3	Defective hepatic mitochondrial respiratory chain in patients with nonalcoholic steatohepatitis. Hepatology, 2003, 38, 999-1007.	7.3	358
4	Mitochondrial Respiration Controls Lysosomal Function during Inflammatory T Cell Responses. Cell Metabolism, 2015, 22, 485-498.	16.2	239
5	Mitochondrial Complex I Plays an Essential Role in Human Respirasome Assembly. Cell Metabolism, 2012, 15, 324-335.	16.2	234
6	Mitochondrial respiratory activity is altered in osteoarthritic human articular chondrocytes. Arthritis and Rheumatism, 2003, 48, 700-708.	6.7	195
7	McArdle disease: what do neurologists need to know?. Nature Clinical Practice Neurology, 2008, 4, 568-577.	2.5	195
8	Complex I Defect in muscle from patients with Huntington's disease. Annals of Neurology, 1998, 43, 397-400.	5.3	154
9	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
10	Mitochondrial respiratory chain dysfunction: Implications in neurodegeneration. Free Radical Biology and Medicine, 2012, 53, 595-609.	2.9	132
11	Effect of nitric oxide on mitochondrial respiratory activity of human articular chondrocytes. Annals of the Rheumatic Diseases, 2004, 64, 388-395.	0.9	122
12	Mitochondrial activity is modulated by TNFÎ± and IL-1Î² in normal human chondrocyte cells. Osteoarthritis and Cartilage, 2006, 14, 1011-1022.	1.3	121
13	X-linkedNDUFA1gene mutations associated with mitochondrial encephalomyopathy. Annals of Neurology, 2007, 61, 73-83.	5.3	118
14	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
15	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
16	Renal pathology in children with mitochondrial diseases. Pediatric Nephrology, 2005, 20, 1299-1305.	1.7	105
17	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
18	Molecular heterogeneity of myophosphorylase deficiency (Mcardle's disease): A genotype-phenotype correlation study. Annals of Neurology, 2001, 50, 574-581.	5.3	86

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19	Favorable Responses to Acute and Chronic Exercise in McArdle Patients. Clinical Journal of Sport Medicine, 2007, 17, 297-303.	1.8	85
20	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
21	Retrospective natural history of thymidine kinase 2 deficiency. Journal of Medical Genetics, 2018, 55, 515-521.	3.2	73
22	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
23	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
24	Secondary coenzyme Q 10 deficiencies in oxidative phosphorylation (OXPHOS) and non-OXPHOS disorders. Mitochondrion, 2016, 30, 51-58.	3.4	70
25	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
26	McArdle Disease: Update of Reported Mutations and Polymorphisms in thePYGMGene. Human Mutation, 2015, 36, 669-678.	2.5	66
27	AÎ² accumulation in choroid plexus is associated with mitochondrial-induced apoptosis. Neurobiology of Aging, 2010, 31, 1569-1581.	3.1	63
28	Cellular pathophysiological consequences of BCS1L mutations in mitochondrial complex III enzyme deficiency. Human Mutation, 2010, 31, 930-941.	2.5	61
29	Biological roles of L-carnitine in perinatal metabolism. Early Human Development, 1998, 53, S43-S50.	1.8	59
30	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
31	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.	2.5	55
32	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
33	Multiple pathways coordinate assembly of human mitochondrial complex IV and stabilization of respiratory supercomplexes. EMBO Journal, 2020, 39, e103912.	7.8	54
34	Mobilisation of mesenchymal cells into blood in response to skeletal muscle injury. British Journal of Sports Medicine, 2006, 40, 719-722.	6.7	53
35	Kearns-Sayre syndrome: Cerebral folate deficiency, MRI findings and new cerebrospinal fluid biochemical features. Mitochondrion, 2010, 10, 429-432.	3.4	53
36	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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37	Myocardial carnitine and carnitine palmitoyltransferase deficiencies in patients with severe heart failure. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2000, 1502, 330-336.	3.8	51
38	Early onset multisystem mitochondrial disorder caused by a nonsense mutation in the mitochondrial DNACytochrome C oxidase IIgene. Annals of Neurology, 2001, 50, 409-413.	5.3	51
39	Thyroid hormone regulates oxidative phosphorylation in the cerebral cortex and striatum of neonatal rats. Journal of Neurochemistry, 2001, 78, 1054-1063.	3.9	50
40	Effect of nitric oxide on mitochondrial activity of human synovial cells. BMC Musculoskeletal Disorders, 2011, 12, 42.	1.9	50
41	Knock-in mice for the R50X mutation in the PYGM gene present with McArdle disease. Brain, 2012, 135, 2048-2057.	7.6	48
42	Prevalence and progression of mitochondrial diseases: A study of 50 patients. Muscle and Nerve, 2003, 28, 690-695.	2.2	44
43	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
44	Genotype Distributions in Top-level Soccer Players: A Role forACE?. International Journal of Sports Medicine, 2009, 30, 387-392.	1.7	43
45	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
46	Genotype modulators of clinical severity in McArdle disease. Neuroscience Letters, 2007, 422, 217-222.	2.1	40
47	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
48	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
49	Nitric oxide compounds have different effects profiles on human articular chondrocyte metabolism. Arthritis Research and Therapy, 2013, 15, R115.	3.5	38
50	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
51	Sporadic MERRF/MELAS overlap syndrome associated with the 3243 tRNALeu(UUR) mutation of mitochondrial DNA. , 1996, 19, 187-190.		36
52	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
53	Impact of the Mitochondrial Genetic Background in Complex III Deficiency. PLoS ONE, 2010, 5, e12801.	2.5	34
54	Molecular analysis of the superoxide dismutase 1 gene in Spanish patients with sporadic or familial amyotrophic lateral sclerosis. Muscle and Nerve, 2002, 26, 274-278.	2.2	32

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55	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
56	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
57	Pathogenic mutations in the 5â€² untranslated region of BCS1L mRNA in mitochondrial complex III deficiency. Mitochondrion, 2009, 9, 299-305.	3.4	29
58	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
59	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
60	Late-onset thymidine kinase 2 deficiency: a review of 18 cases. Orphanet Journal of Rare Diseases, 2019, 14, 100.	2.7	29
61	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
62	Regulation of Mitochondrial Function by the Actin Cytoskeleton. Frontiers in Cell and Developmental Biology, 2021, 9, 795838.	3.7	28
63	Identification of novel mutations in Spanish patients with muscle carnitine palmitoyltransferase II deficiency. Human Mutation, 2000, 15, 579-580.	2.5	27
64	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
65	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
66	Molecular Analysis of Myophosphorylase Deficiency in Dutch Patients with McArdle's Disease. Annals of Human Genetics, 2004, 68, 17-22.	0.8	26
67	Severe TK2 enzyme activity deficiency in patients with mild forms of myopathy. Neurology, 2015, 84, 2286-2288.	1.1	26
68	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
69	Coenzyme Q10 deficiency associated with a mitochondrial DNA depletion syndrome: A case report. Clinical Biochemistry, 2009, 42, 742-745.	1.9	25
70	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
71	Association of genetically proven deficiencies of myophosphorylase and AMP deaminase: a second case of â€˜double troubleâ€™. Neuromuscular Disorders, 1997, 7, 387-389.	0.6	23
72	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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73	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
74	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
75	A Transcriptomic Approach to Search for Novel Phenotypic Regulators in McArdle Disease. PLoS ONE, 2012, 7, e31718.	2.5	22
76	Are mitochondrial haplogroups associated with extreme longevity? A study on a Spanish cohort. Age, 2012, 34, 227-233.	3.0	22
77	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
78	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
79	Differential proteomic profiling unveils new molecular mechanisms associated with mitochondrial complex III deficiency. Journal of Proteomics, 2015, 113, 38-56.	2.4	21
80	Clinical, pathological and genetic spectrum in 89 cases of mitochondrial progressive external ophthalmoplegia. Journal of Medical Genetics, 2020, 57, 643-646.	3.2	21
81	Novel mutations in patients with McArdle disease by analysis of skeletal muscle mRNA. Journal of Medical Genetics, 2008, 46, 198-202.	3.2	20
82	Are mitochondrial haplogroups associated with elite athletic status? A study on a Spanish cohort. Mitochondrion, 2011, 11, 905-908.	3.4	20
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	Growth Differentiation Factor 15 is a potential biomarker of therapeutic response for TK2 deficient myopathy. Scientific Reports, 2020, 10, 10111.	3.3	20
85	Can patients with McArdle's disease run? * Commentary. British Journal of Sports Medicine, 2006, 41, 53-54.	6.7	19
86	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
87	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
88	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
89	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.	3.6	18
90	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

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91	Collagen XIX Alpha 1 Improves Prognosis in Amyotrophic Lateral Sclerosis. , 2019, 10, 278.		18
92	Apolipoprotein E polymorphism and carotid atherosclerosis in patients with coronary disease. International Journal of Cardiology, 2004, 94, 209-212.	1.7	17
93	Excessive skeletal muscle recruitment during strenuous exercise in McArdle patients. European Journal of Applied Physiology, 2010, 110, 1047-1055.	2.5	17
94	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
95	Phenotype consequences of myophosphorylase dysfunction: insights from the McArdle mouse model. Journal of Physiology, 2015, 593, 2693-2706.	2.9	17
96	Functional Characterization of Three Concomitant MtDNA LHON Mutations Shows No Synergistic Effect on Mitochondrial Activity. PLoS ONE, 2016, 11, e0146816.	2.5	17
97	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
98	Congenital neurogenic muscular atrophy in megaconial myopathy due to a mutation in CHKB gene. Brain and Development, 2016, 38, 167-172.	1.1	16
99	Health Benefits of an Innovative Exercise Program for Mitochondrial Disorders. Medicine and Science in Sports and Exercise, 2018, 50, 1142-1151.	0.4	16
100	Single large-scale mitochondrial DNA deletion in a patient with encephalopathy, cardiomyopathy, and prominent intestinal pseudo-obstruction. Neuromuscular Disorders, 2000, 10, 56-58.	0.6	15
101	A new mtDNA mutation in the tRNALeu(UUR) gene associated with ocular myopathy. Neuromuscular Disorders, 2001, 11, 477-480.	0.6	15
102	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.	2.0	15
103	The A8296G mtDNA mutation associated with several mitochondrial diseases does not cause mitochondrial dysfunction in cybrid cell lines. Human Mutation, 2002, 19, 234-239.	2.5	15
104	C34T mutation of the AMPD1 gene in an elite white runner. British Journal of Sports Medicine, 2006, 40, e7-e7.	6.7	15
105	Exercise Capacity in a Child With McArdle Disease. Journal of Child Neurology, 2007, 22, 880-882.	1.4	15
106	Mitochondrial tRNA valine as a recurrent target for mutations involved in mitochondrial cardiomyopathies. Mitochondrion, 2012, 12, 357-362.	3.4	15
107	Genes and exercise intolerance: insights from McArdle disease. Physiological Genomics, 2016, 48, 93-100.	2.3	15
108	Physical Exercise and Mitochondrial Disease: Insights From a Mouse Model. Frontiers in Neurology, 2019, 10, 790.	2.4	15

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109	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
110	Two pathogenic mutations in the mitochondrial DNA tRNA Leu(UUR) gene (T3258C and A3280G) resulting in variable clinical phenotypes. Neuromuscular Disorders, 2003, 13, 416-420.	0.6	14
111	Mild ocular myopathy associated with a novel mutation in mitochondrial twinkle helicase. Neuromuscular Disorders, 2007, 17, 677-680.	0.6	14
112	Increased dNTP pools rescue mtDNA depletion in human POLGâ€deficient fibroblasts. FASEB Journal, 2019, 33, 7168-7179.	0.5	14
113	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
114	Next-generation sequencing to estimate the prevalence of a great unknown: McArdle disease. Genetics in Medicine, 2015, 17, 679-680.	2.4	13
115	Muscle Signaling in Exercise Intolerance. Medicine and Science in Sports and Exercise, 2016, 48, 1448-1458.	0.4	13
116	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
117	Missense mutations have unexpected consequences: The McArdle disease paradigm. Human Mutation, 2018, 39, 1338-1343.	2.5	13
118	Genes and Variants Underlying Human Congenital Lactic Acidosisâ€"From Genetics to Personalized Treatment. Journal of Clinical Medicine, 2019, 8, 1811.	2.4	13
119	Preclinical Research in Glycogen Storage Diseases: A Comprehensive Review of Current Animal Models. International Journal of Molecular Sciences, 2020, 21, 9621.	4.1	13
120	Cosegregation of the mitochondrial DNA A1555G and G4309A mutations results in deafness and mitochondrial myopathy. Muscle and Nerve, 2002, 25, 185-188.	2.2	12
121	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
122	Cerebrospinal fluid carnitine levels in patients with Parkinson's disease. Journal of the Neurological Sciences, 1997, 145, 183-185.	0.6	11
123	A mitochondrial tRNALys gene mutation (T8316C) in a patient with mitochondrial myopathy, lactic acidosis, and stroke-like episodes. Neuromuscular Disorders, 2000, 10, 493-496.	0.6	11
124	AMPD1 Genotypes and Exercise Capacity in McArdle Patients. International Journal of Sports Medicine, 2008, 29, 331-335.	1.7	11
125	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
126	Low survival rate and muscle fiber-dependent aging effects in the McArdle disease mouse model. Scientific Reports, 2019, 9, 5116.	3.3	11

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127	Altered Expression Ratio of Actin-Binding Gelsolin Isoforms Is a Novel Hallmark of Mitochondrial OXPHOS Dysfunction. Cells, 2020, 9, 1922.	4.1	11
128	Chronic primary intestinal pseudo-obstruction from visceral myopathy. Revista Espanola De Enfermedades Digestivas, 2006, 98, 292-302.	0.3	11
129	Abnormal carnitine distribution in the muscles of patients with idiopathic inflammatory myopathy. Arthritis and Rheumatism, 1996, 39, 1869-1874.	6.7	10
130	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
131	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
132	Small GTPases of the Ras superfamily and glycogen phosphorylase regulation in T cells. Small GTPases, 2021, 12, 106-113.	1.6	10
133	Plasma LDH: A specific biomarker for lung affectation in COVID-19?. Practical Laboratory Medicine, 2021, 25, e00226.	1.3	10
134	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
135	A missense mutation T487N in the myophosphorylase gene in a Spanish patient with McArdle's disease. Neuromuscular Disorders, 2000, 10, 138-140.	0.6	9
136	A homozygous missense mutation (A659D) in the myophosphorylase gene in a Spanish patient with McArdle's disease. Neuromuscular Disorders, 2000, 10, 447-449.	0.6	9
137	Two novel mutations in the muscle glycogen phosphorylase gene in McArdle's disease. Muscle and Nerve, 2003, 28, 380-382.	2.2	9
138	Novel Mutation in the PYGM Gene Resulting in McArdle Disease. Archives of Neurology, 2006, 63, 1782.	4.5	9
139	McArdle disease: Another systemic low-inflammation disorder?. Neuroscience Letters, 2008, 431, 106-111.	2.1	9
140	Mitochondrial Disorders Due to Nuclear OXPHOS Gene Defects. Advances in Experimental Medicine and Biology, 2009, 652, 85-116.	1.6	9
141	Primary Adenosine Monophosphate (AMP) Deaminase Deficiency in a Hypotonic Infant. Journal of Child Neurology, 2011, 26, 734-737.	1.4	9
142	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
143	A New Condition in McArdle Disease. Medicine and Science in Sports and Exercise, 2018, 50, 3-10.	0.4	9
144	Deoxynucleoside therapy for respiratory involvement in adult patients with thymidine kinase 2-deficient myopathy. BMJ Open Respiratory Research, 2020, 7, e000774.	3.0	9

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145	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
146	McArdle disease does not affect skeletal muscle fibre type profiles in humans. Biology Open, 2014, 3, 1224-1227.	1.2	8
147	Assessment of resting energy expenditure in pediatric mitochondrial diseases with indirect calorimetry. Clinical Nutrition, 2016, 35, 1484-1489.	5.0	8
148	The homozygous R504C mutation in MTO1 gene is responsible for ONCE syndrome. Clinical Genetics, 2017, 91, 46-53.	2.0	8
149	Novel NDUFA13 Mutations Associated with OXPHOS Deficiency and Leigh Syndrome: A Second Family Report. Genes, 2020, 11, 855.	2.4	8
150	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
151	C34T mutation of the AMPD1 gene in an elite white runner. BMJ Case Reports, 2009, 2009, bcr0720080535-bcr0720080535.	0.5	8
152	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
153	The Genetic Landscape of Mitochondrial Diseases in Spain: A Nationwide Call. Genes, 2021, 12, 1590.	2.4	8
154	The V368i mutation in Twinkle does not segregate with adPEO. Annals of Neurology, 2003, 53, 278-278.	5.3	7
155	A novel RRM2B gene variant associated with Telbivudine-induced mitochondrial myopathy. Journal of the Neurological Sciences, 2015, 358, 481-483.	0.6	7
156	First missense mutation outside of SERAC1 lipase domain affecting intracellular cholesterol trafficking. Neurogenetics, 2016, 17, 51-56.	1.4	7
157	Myopathic mtDNA Depletion Syndrome Due to Mutation in TK2 Gene. Pediatric and Developmental Pathology, 2017, 20, 416-420.	1.0	7
158	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
159	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
160	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
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	Polymorphisms influencing muscle phenotypes in North-African and Spanish populations. Annals of Human Biology, 2012, 39, 166-169.	1.0	6

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163	211th ENMC International Workshop:. Neuromuscular Disorders, 2017, 27, 1143-1151.	0.6	6
164	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
165	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
166	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
167	Cardiac Dysfunction in Mitochondrial Disease. Circulation Journal, 2013, 77, 2799-2806.	1.6	5
168	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
169	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
170	When should a nephrologist suspect a mitochondrial disease?. Nefrologia, 2015, 35, 6-17.	0.4	5
171	Establishment of a human iPSC line (IISHDOi001-A) from a patient with McArdle disease. Stem Cell Research, 2017, 23, 188-192.	0.7	4
172	Muscle fiber type proportion and size is not altered in mcardle disease. Muscle and Nerve, 2017, 55, 916-918.	2.2	4
173	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
174	Absence of p.R50X Pygm read-through in McArdle disease cellular models. DMM Disease Models and Mechanisms, 2019, 13, .	2.4	4
175	Exercise Training and Neurodegeneration in Mitochondrial Disorders: Insights From the Harlequin Mouse. Frontiers in Physiology, 2020, 11, 594223.	2.8	4
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