Michio Hirano

List of Publications by Year
in descending order

229 papers	19,655 citations	7087 78 h-index	12258 133 g-index
258 all docs	258 docs citations	258 times ranked	17167 citing authors

#	Article	IF	Citations
1	Mitochondrial diseases. Nature Reviews Disease Primers, 2016, 2, 16080.	18.1	1,001
2	Primary LAMP-2 deficiency causes X-linked vacuolar cardiomyopathy and myopathy (Danon disease). Nature, 2000, 406, 906-910.	13.7	865
3	Thymidine Phosphorylase Gene Mutations in MNGIE, a Human Mitochondrial Disorder. Science, 1999, 283, 689-692.	6.0	827
4	Human mitochondrial DNA: roles of inherited and somatic mutations. Nature Reviews Genetics, 2012, 13, 878-890.	7.7	620
5	Fatal infantile cardioencephalomyopathy with COX deficiency and mutations in SCO2, a COX assembly gene. Nature Genetics, 1999, 23, 333-337.	9.4	556
6	Diagnosis and management of mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. Genetics in Medicine, 2015, 17, 689-701.	1.1	414
7	Leigh Syndrome with Nephropathy and CoQ10 Deficiency Due to decaprenyl diphosphate synthase subunit 2 (PDSS2) Mutations. American Journal of Human Genetics, 2006, 79, 1125-1129.	2.6	359
8	FGF-21 as a biomarker for muscle-manifesting mitochondrial respiratory chain deficiencies: a diagnostic study. Lancet Neurology, The, 2011, 10, 806-818.	4.9	352
9	Topical Review: Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, and Strokelike Episodes (MELAS): Current Concepts. Journal of Child Neurology, 1994, 9, 4-13.	0.7	351
10	MELAS: An original case and clinical criteria for diagnosis. Neuromuscular Disorders, 1992, 2, 125-135.	0.3	330
11	Mitochondrial neurogastrointestinal encephalomyopathy: An autosomal recessive disorder due to thymidine phosphorylase mutations. Annals of Neurology, 2000, 47, 792-800.	2.8	324
12	A Mutation in Para-Hydroxybenzoate-Polyprenyl Transferase (COQ2) Causes Primary Coenzyme Q10 Deficiency. American Journal of Human Genetics, 2006, 78, 345-349.	2.6	322
13	The myopathic form of coenzyme Q10 deficiency is caused by mutations in the electron-transferring-flavoprotein dehydrogenase (ETFDH) gene. Brain, 2007, 130, 2037-2044.	3.7	298
14	The clinical maze of mitochondrial neurology. Nature Reviews Neurology, 2013, 9, 429-444.	4.9	293
15	ADCK3, an Ancestral Kinase, Is Mutated in a Form of Recessive Ataxia Associated with Coenzyme Q10 Deficiency. American Journal of Human Genetics, 2008, 82, 661-672.	2.6	290
16	Complex I deficiency primes Bax-dependent neuronal apoptosis through mitochondrial oxidative damage. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 19126-19131.	3.3	273
17	Novel cell lines derived from adult human ventricular cardiomyocytes. Journal of Molecular and Cellular Cardiology, 2005, 39, 133-147.	0.9	236
18	Nuclear genome transfer in human oocytes eliminates mitochondrial DNA variants. Nature, 2013, 493, 632-637.	13.7	223

#	Article	IF	Citations
19	Atypical clinical presentations associated with the MELAS mutation at position 3243 of human mitochondrial DNA. Neuromuscular Disorders, 1993, 3, 43-50.	0.3	219
20	Altered Thymidine Metabolism Due to Defects of Thymidine Phosphorylase. Journal of Biological Chemistry, 2002, 277, 4128-4133.	1.6	209
21	A Nonsense Mutation in COQ9 Causes Autosomal-Recessive Neonatal-Onset Primary Coenzyme Q10 Deficiency: A Potentially Treatable Form of Mitochondrial Disease. American Journal of Human Genetics, 2009, 84, 558-566.	2.6	206
22	GD3 ganglioside is a glycolipid characteristic of immature neuroectodermal cells. Journal of Neuroimmunology, 1984, 7, 179-192.	1.1	193
23	Heterogeneity of Coenzyme Q₁₀Deficiency. Archives of Neurology, 2012, 69, 978-83.	4.9	192
24	Clinical and genetic spectrum of mitochondrial neurogastrointestinal encephalomyopathy. Brain, 2011, 134, 3326-3332.	3.7	191
25	Genetic Drift Can Compromise Mitochondrial Replacement by Nuclear Transfer in Human Oocytes. Cell Stem Cell, 2016, 18, 749-754.	5.2	170
26	Site-specific somatic mitochondrial DNA point mutations in patients with thymidine phosphorylase deficiency. Journal of Clinical Investigation, 2003, 111, 1913-1921.	3.9	165
27	Human Coenzyme Q10 Deficiency. Neurochemical Research, 2007, 32, 723-727.	1.6	163
28	A homoplasmic mitochondrial transfer Ribonucleic Acid mutation as a cause of maternally inherited hypertrophic cardiomyopathy. Journal of the American College of Cardiology, 2003, 41, 1786-1796.	1.2	161
29	Navajo Neurohepatopathy Is Caused by a Mutation in the MPV17 Gene. American Journal of Human Genetics, 2006, 79, 544-548.	2.6	158
30	Coenzyme Q and mitochondrial disease. Developmental Disabilities Research Reviews, 2010, 16, 183-188.	2.9	157
31	New treatments for mitochondrial diseaseâ€"no time to drop our standards. Nature Reviews Neurology, 2013, 9, 474-481.	4.9	157
32	Mitochondrial Neurogastrointestinal Encephalomyopathy (MNGIE): A Disease of Two Genomes. Neurologist, 2004, 10, 8-17.	0.4	156
33	Mitochondrial DNA depletion anddGKgene mutations. Annals of Neurology, 2002, 52, 311-317.	2.8	152
34	Respiratory chain dysfunction and oxidative stress correlate with severity of primary CoQ ₁₀ deficiency. FASEB Journal, 2008, 22, 1874-1885.	0.2	150
35	Recurrent De Novo and Biallelic Variation of ATAD3A , Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. American Journal of Human Genetics, 2016, 99, 831-845.	2.6	146
36	Reactive oxygen species, oxidative stress, and cell death correlate with level of CoQ ₁₀ deficiency. FASEB Journal, 2010, 24, 3733-3743.	0.2	142

#	Article	IF	Citations
37	Apparent mtDNA heteroplasmy in Alzheimer's disease patients and in normals due to PCR amplification of nucleus-embedded mtDNA pseudogenes. Proceedings of the National Academy of Sciences of the United States of America, 1997, 94, 14894-14899.	3.3	137
38	Mitochondria in neuromuscular disorders1This paper is dedicated to the memory of Giovanni Salviati, a great scientist and a dear friend.1. Biochimica Et Biophysica Acta - Bioenergetics, 1998, 1366, 199-210.	0.5	136
39	POLG mutations and Alpers syndrome. Annals of Neurology, 2005, 57, 921-923.	2.8	131
40	Mitochondrial encephalomyopathies: an update. Neuromuscular Disorders, 2005, 15, 276-286.	0.3	130
41	Approaches to the treatment of mitochondrial diseases. Muscle and Nerve, 2006, 34, 265-283.	1.0	130
42	Missense mutation of the COQ2 gene causes defects of bioenergetics and de novo pyrimidine synthesis. Human Molecular Genetics, 2007, 16, 1091-1097.	1.4	129
43	A Diagnostic Algorithm for Metabolic Myopathies. Current Neurology and Neuroscience Reports, 2010, 10, 118-126.	2.0	128
44	Allogeneic haematopoietic stem cell transplantation for mitochondrial neurogastrointestinal encephalomyopathy. Brain, 2015, 138, 2847-2858.	3.7	128
45	Does Linezolid Cause Lactic Acidosis by Inhibiting Mitochondrial Protein Synthesis?. Clinical Infectious Diseases, 2005, 40, e113-e116.	2.9	123
46	Unbalanced deoxynucleotide pools cause mitochondrial DNA instability in thymidine phosphorylase-deficient mice. Human Molecular Genetics, 2009, 18, 714-722.	1.4	123
47	ETFDH mutations, CoQ10 levels, and respiratory chain activities in patients with riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency. Neuromuscular Disorders, 2009, 19, 212-216.	0.3	118
48	The G13513A Mutation in the ND5 Gene of Mitochondrial DNA as a Common Cause of MELAS or Leigh Syndrome. Archives of Neurology, 2008, 65, 368-72.	4.9	113
49	Emerging therapies for mitochondrial diseases. Essays in Biochemistry, 2018, 62, 467-481.	2.1	113
50	Glycogen branching enzyme deficiency in adult polyglucosan body disease. Annals of Neurology, 1993, 33, 88-93.	2.8	112
51	Molecular basis of infantile reversible cytochrome c oxidase deficiency myopathy. Brain, 2009, 132, 3165-3174.	3.7	112
52	Human CoQ₁₀ deficiencies. BioFactors, 2008, 32, 113-118.	2.6	110
53	X-Linked Dominant Scapuloperoneal Myopathy Is Due to a Mutation in the Gene Encoding Four-and-a-Half-LIM Protein 1. American Journal of Human Genetics, 2008, 82, 208-213.	2.6	108
54	Maternally Inherited Cardiomyopathy: An Atypical Presentation of the mtDNA 12S rRNA Gene A1555G Mutation. American Journal of Human Genetics, 1999, 64, 295-300.	2.6	107

#	Article	IF	Citations
55	Definitive Diagnosis of Mitochondrial Neurogastrointestinal Encephalomyopathy by Biochemical Assays. Clinical Chemistry, 2004, 50, 120-124.	1.5	107
56	Defects of intergenomic communication: autosomal disorders that cause multiple deletions and depletion of mitochondrial DNA. Seminars in Cell and Developmental Biology, 2001, 12, 417-427.	2.3	105
57	Clinical and Genetic Heterogeneity in Progressive External Ophthalmoplegia Due to Mutations in Polymerase Î³. Archives of Neurology, 2003, 60, 1279-84.	4.9	104
58	A functionally dominant mitochondrial DNA mutation. Human Molecular Genetics, 2008, 17, 1814-1820.	1.4	104
59	Thymidine kinase 2 (H126N) knockin mice show the essential role of balanced deoxynucleotide pools for mitochondrial DNA maintenance. Human Molecular Genetics, 2008, 17, 2433-2440.	1.4	101
60	Gastrointestinal Dysmotility in Mitochondrial Neurogastrointestinal Encephalomyopathy Is Caused by Mitochondrial DNA Depletion. American Journal of Pathology, 2008, 173, 1120-1128.	1.9	100
61	Therapeutic prospects for mitochondrial disease. Trends in Molecular Medicine, 2010, 16, 268-276.	3.5	97
62	Mitochondrial encephalomyopathies: Clinical and molecular analysis. Journal of Bioenergetics and Biomembranes, 1994, 26, 291-299.	1.0	96
63	Primary and secondary CoQ₁₀ deficiencies in humans. BioFactors, 2011, 37, 361-365.	2.6	96
64	Haploinsufficiency ofCOQ4causes coenzyme Q₁₀deficiency. Journal of Medical Genetics, 2012, 49, 187-191.	1.5	95
65	Circulating markers of NADH-reductive stress correlate with mitochondrial disease severity. Journal of Clinical Investigation, 2021, 131, .	3.9	95
66	Treatment of CoQ10 Deficient Fibroblasts with Ubiquinone, CoQ Analogs, and Vitamin C: Time- and Compound-Dependent Effects. PLoS ONE, 2010, 5, e11897.	1.1	92
67	Diagnostic odyssey of patients with mitochondrial disease. Neurology: Genetics, 2018, 4, e230.	0.9	92
68	Mitochondrial Neurogastrointestinal Encephalomyopathy Syndrome Maps to Chromosome 22q13.32-qter. American Journal of Human Genetics, 1998, 63, 526-533.	2.6	91
69	Elevated plasma deoxyuridine in patients with thymidine phosphorylase deficiency. Biochemical and Biophysical Research Communications, 2003, 303, 14-18.	1.0	91
70	Deoxycytidine and Deoxythymidine Treatment for Thymidine Kinase 2 Deficiency. Annals of Neurology, 2017, 81, 641-652.	2.8	89
71	Mutations in coenzyme Q10 biosynthetic genes. Journal of Clinical Investigation, 2007, 117, 587-589.	3.9	89
72	ND5 is a hot-spot for multiple atypical mitochondrial DNA deletions in mitochondrial neurogastrointestinal encephalomyopathy. Human Molecular Genetics, 2003, 13, 91-101.	1.4	88

#	Article	IF	Citations
73	Mitochondrial DNA depletion syndrome due to mutations in the RRM2B gene. Neuromuscular Disorders, 2008, 18, 453-459.	0.3	87
74	Maintenance of Human Rearranged Mitochondrial DNAs in Long-Term Cultured Transmitochondrial Cell Lines. Molecular Biology of the Cell, 2000, 11, 2349-2358.	0.9	85
75	Jumping Performance of Frogs (Rana Pipiens) as a Function of Muscle Temperature. Journal of Experimental Biology, 1984, 108, 429-439.	0.8	85
76	A <scp>POGLUT</scp> 1 mutation causes a muscular dystrophy with reduced Notch signaling and satellite cell loss. EMBO Molecular Medicine, 2016, 8, 1289-1309.	3.3	84
77	CoQ10 deficiencies and MNGIE: Two treatable mitochondrial disorders. Biochimica Et Biophysica Acta - General Subjects, 2012, 1820, 625-631.	1.1	83
78	Inhibition of NADPH oxidase 2 (NOX2) prevents sepsis-induced cardiomyopathy by improving calcium handling and mitochondrial function. JCI Insight, 2017, 2, .	2.3	83
79	Autophagic Vacuoles with Sarcolemmal Features Delineate Danon Disease and Related Myopathies. Journal of Neuropathology and Experimental Neurology, 2005, 64, 513-522.	0.9	81
80	Senataxin mutations and amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2011, 12, 223-227.	2.3	81
81	Deoxypyrimidine monophosphate bypass therapy for thymidine kinase 2 deficiency. EMBO Molecular Medicine, 2014, 6, 1016-1027.	3.3	79
82	Inhibition of NAPDH Oxidase 2 (NOX2) Prevents Oxidative Stress and Mitochondrial Abnormalities Caused by Saturated Fat in Cardiomyocytes. PLoS ONE, 2016, 11, e0145750.	1.1	78
83	Identical Mitochondrial DNA Deletion in a Woman with Ocular Myopathy and in Her Son with Pearson Syndrome. American Journal of Human Genetics, 2002, 71, 679-683.	2.6	76
84	Mitochondrial DNA and RNA processing in MELAS. Annals of Neurology, 1996, 40, 172-180.	2.8	74
85	Clinical and genetic analysis of lipid storage myopathies. Muscle and Nerve, 2009, 39, 333-342.	1.0	74
86	Retrospective natural history of thymidine kinase 2 deficiency. Journal of Medical Genetics, 2018, 55, 515-521.	1.5	73
87	Deoxynucleoside Therapy for Thymidine Kinase 2â€“Deficient Myopathy. Annals of Neurology, 2019, 86, 293-303.	2.8	72
88	Natural underlying mt<scp>DNA</scp> heteroplasmy as a potential source of intraâ€person hi<scp>PSC</scp> variability. EMBO Journal, 2016, 35, 1979-1990.	3.5	71
89	Low-dose rapamycin extends lifespan in a mouse model of mtDNA depletion syndrome. Human Molecular Genetics, 2017, 26, 4588-4605.	1.4	70
90	Mitochondrial DNA Depletion and Thymidine Phosphate Pool Dynamics in a Cellular Model of Mitochondrial Neurogastrointestinal Encephalomyopathy. Journal of Biological Chemistry, 2006, 281, 22720-22728.	1.6	68

#	Article	IF	Citations
91	MPV17 Mutations Causing Adult-Onset Multisystemic Disorder With Multiple Mitochondrial DNA Deletions. Archives of Neurology, 2012, 69, 1648.	4.9	68
92	Administration of deoxyribonucleosides or inhibition of their catabolism as a pharmacological approach for mitochondrial DNA depletion syndrome. Human Molecular Genetics, 2014, 23, 2459-2467.	1.4	67
93	MPV17 Loss Causes Deoxynucleotide Insufficiency and Slow DNA Replication in Mitochondria. PLoS Genetics, 2016, 12, e1005779.	1.5	67
94	Mitochondria and the heart. Current Opinion in Cardiology, 2001, 16, 201-210.	0.8	65
95	Association of myopathy with large-scale mitochondrial dna duplications and deletions: Which is pathogenic?. Annals of Neurology, 1997, 42, 180-188.	2.8	64
96	MNGIE: from nuclear DNA to mitochondrial DNA. Neuromuscular Disorders, 2001, 11, 7-10.	0.3	64
97	Late-onset MNGIE due to partial loss of thymidine phosphorylase activity. Annals of Neurology, 2005, 58, 649-652.	2.8	64
98	Thymidine and deoxyuridine accumulate in tissues of patients with mitochondrial neurogastrointestinal encephalomyopathy (MNGIE). FEBS Letters, 2007, 581, 3410-3414.	1.3	64
99	Mitochondrial Neurogastrointestinal Encephalomyopathy: Evidence of Mitochondrial DNA Depletion in the Small Intestine. Gastroenterology, 2006, 130, 893-901.	0.6	63
100	Tissueâ€specific oxidative stress and loss of mitochondria in CoQâ€deficient Pdss2 mutant mice. FASEB Journal, 2013, 27, 612-621.	0.2	61
101	Three-Dimensional Analysis of Mitochondrial Crista Ultrastructure in a Patient with Leigh Syndrome by In Situ Cryoelectron Tomography. IScience, 2018, 6, 83-91.	1.9	60
102	Neutral lipid storage disease with subclinical myopathy due to a retrotransposal insertion in the PNPLA2 gene. Neuromuscular Disorders, 2010, 20, 397-402.	0.3	58
103	International Workshop:. Neuromuscular Disorders, 2017, 27, 1126-1137.	0.3	58
104	Mitochondrial Myopathy of Childhood Associated With Mitochondrial DNA Depletion and a Homozygous Mutation (T77M) in the TK2 Gene. Archives of Neurology, 2003, 60, 1007.	4.9	57
105	Gene Therapy Using a Liver-targeted AAV Vector Restores Nucleoside and Nucleotide Homeostasis in a Murine Model of MNGIE. Molecular Therapy, 2014, 22, 901-907.	3.7	55
106	Protean Phenotypic Features of the A3243G Mitochondrial DNA Mutation. Archives of Neurology, 2009, 66, 85-91.	4.9	53
107	TheCOQ2genotype predicts the severity of coenzyme Q₁₀deficiency. Human Molecular Genetics, 2016, 25, 4256-4265.	1.4	53
108	A novel mitochondrial tRNALeu(UUR) mutation in a patient with features of MERRF and Kearnsâ€“Sayre syndrome. Neuromuscular Disorders, 2003, 13, 334-340.	0.3	51

#	Article	IF	Citations
109	Defects of Intergenomic Communication: Where Do We Stand?. Brain Pathology, 2000, 10, 451-461.	2.1	50
110	Primary coenzyme Q₁₀ deficiency and the brain. BioFactors, 2003, 18, 145-152.	2.6	49
111	Thymidine phosphorylase mutations cause instability of mitochondrial DNA. Gene, 2005, 354, 152-156.	1.0	49
112	Functional characterization of human COQ4, a gene required for Coenzyme Q10 biosynthesis. Biochemical and Biophysical Research Communications, 2008, 372, 35-39.	1.0	49
113	Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE): Position paper on diagnosis, prognosis, and treatment by the <scp>MNGIE</scp> International Network. Journal of Inherited Metabolic Disease, 2021, 44, 376-387.	1.7	47
114	USMG5 Ashkenazi Jewish founder mutation impairs mitochondrial complex V dimerization and ATP synthesis. Human Molecular Genetics, 2018, 27, 3305-3312.	1.4	45
115	Branching Enzyme Deficiency. JAMA Neurology, 2014, 71, 41.	4.5	43
116	Nutritional interventions in primary mitochondrial disorders: Developing an evidence base. Molecular Genetics and Metabolism, 2016, 119, 187-206.	0.5	41
117	Effects of Inhibiting CoQ10 Biosynthesis with 4-nitrobenzoate in Human Fibroblasts. PLoS ONE, 2012, 7, e30606.	1.1	40
118	Long-Term Sustained Effect of Liver-Targeted Adeno-Associated Virus Gene Therapy for Mitochondrial Neurogastrointestinal Encephalomyopathy. Human Gene Therapy, 2018, 29, 708-718.	1.4	39
119	Clinical Presentations of Coenzyme Q10 Deficiency Syndrome. Molecular Syndromology, 2014, 5, 141-146.	0.3	38
120	Mitochondrial diseases in North America. Neurology: Genetics, 2020, 6, e402.	0.9	38
121	MRI of Five Patients with Mitochondrial Neurogastrointestinal Encephalomyopathy. American Journal of Roentgenology, 2004, 182, 1537-1541.	1.0	37
122	Thymidine Phosphorylase Participates in Platelet Signaling and Promotes Thrombosis. Circulation Research, 2014, 115, 997-1006.	2.0	37
123	Human aging DNA methylation signatures are conserved but accelerated in cultured fibroblasts. Epigenetics, 2019, 14, 961-976.	1.3	36
124	SETX (senataxin), the helicase mutated in AOA2 and ALS4, functions in autophagy regulation. Autophagy, 2021, 17, 1889-1906.	4.3	34
125	Characterization of Danon disease in a male patient and his affected mother. Neuromuscular Disorders, 2003, 13, 708-711.	0.3	33
126	Advances in primary mitochondrial myopathies. Current Opinion in Neurology, 2019, 32, 715-721.	1.8	32

#	Article	IF	Citations
127	Intracellular calcium leak as a therapeutic target for RYR1-related myopathies. Acta Neuropathologica, 2020, 139, 1089-1104.	3.9	32
128	Pathogenesis and Treatment of Mitochondrial Disorders. Advances in Experimental Medicine and Biology, 2009, 652, 139-170.	0.8	31
129	Pathogenesis of the deafness-associated A1555G mitochondrial DNA mutation. Biochemical and Biophysical Research Communications, 2002, 293, 521-529.	1.0	30
130	Onset and organ specificity of Tk2 deficiency depends on Tk1 down-regulation and transcriptional compensation. Human Molecular Genetics, 2011, 20, 155-164.	1.4	30
131	The clinical spectrum of the congenital myasthenic syndrome resulting from COL13A1 mutations. Brain, 2019, 142, 1547-1560.	3.7	30
132	Mitochondrial diseases. Neurologic Clinics, 2002, 20, 809-839.	0.8	29
133	A novel complex neurological phenotype due to a homozygous mutation in FDX2. Brain, 2018, 141, 2289-2298.	3.7	29
134	Late-onset thymidine kinase 2 deficiency: a review of 18 cases. Orphanet Journal of Rare Diseases, 2019, 14, 100.	1.2	29
135	Epidemic optic and peripheral neuropathy in Cuba: A unique geopolitical public health problem. Survey of Ophthalmology, 1997, 41, 341-353.	1.7	28
136	Amyotrophic Lateral Sclerosis With Ragged-Red Fibers. Archives of Neurology, 2008, 65, 403-6.	4.9	28
137	TK2 mutation presenting as indolent myopathy. Neurology, 2013, 80, 504-506.	1.5	28
138	Long-Term Restoration of Thymidine Phosphorylase Function and Nucleoside Homeostasis Using Hematopoietic Gene Therapy in a Murine Model of Mitochondrial Neurogastrointestinal Encephalomyopathy. Human Gene Therapy, 2016, 27, 656-667.	1.4	26
139	Successful liver transplantation in mitochondrial neurogastrointestinal encephalomyopathy (MNGIE). Molecular Genetics and Metabolism, 2020, 130, 58-64.	0.5	26
140	Metabolic shift underlies recovery in reversible infantile respiratory chain deficiency. EMBO Journal, 2020, 39, e105364.	3.5	26
141	Alteration of Nucleotide Metabolism: A New Mechanism for Mitochondrial Disorders. Clinical Chemistry and Laboratory Medicine, 2003, 41, 845-51.	1.4	25
142	Dystonia-Ataxia with early handwriting deterioration in COQ8A mutation carriers: A case series and literature review. Parkinsonism and Related Disorders, 2019, 68, 8-16.	1.1	25
143	Mitochondrial disease patients' perception of dietary supplements' use. Molecular Genetics and Metabolism, 2016, 119, 100-108.	0.5	24
144	MERRF and Kearnsâ€“Sayre overlap syndrome due to the mitochondrial DNA m.3291T>C mutation. Muscle and Nerve, 2011, 44, 448-451.	1.0	23

#	Article	IF	Citations
145	Pathomechanisms in Coenzyme Q₁₀-Deficient Human Fibroblasts. Molecular Syndromology, 2014, 5, 163-169.	0.3	23
146	Leber's hereditary optic neuropathy mitochondrial DNA mutations in normal-tension glaucoma. Graefe's Archive for Clinical and Experimental Ophthalmology, 2001, 239, 437-440.	1.0	22
147	Human mitochondrial pyrophosphatase: cDNA cloning and analysis of the gene in patients with mtDNA depletion syndromes. Genomics, 2006, 87, 410-416.	1.3	22
148	<scp>GGPS1</scp> Mutations Cause Muscular Dystrophy/Hearing Loss/Ovarian Insufficiency Syndrome. Annals of Neurology, 2020, 88, 332-347.	2.8	22
149	A novel ECGF1 mutation in a Thai patient with mitochondrial neurogastrointestinal encephalomyopathy (MNGIE). Clinical Neurology and Neurosurgery, 2007, 109, 613-616.	0.6	21
150	Coenzyme Q10 Deficiencies in Neuromuscular Diseases. Advances in Experimental Medicine and Biology, 2009, 652, 117-128.	0.8	21
151	Detection of uniparental isodisomy in autosomal recessive mitochondrial DNA depletion syndrome by high-density SNP array analysis. Journal of Human Genetics, 2011, 56, 834-839.	1.1	21
152	Whole exome sequencing identifies a homozygous POLG2 missense variant in an infant with fulminant hepatic failure and mitochondrial DNA depletion. European Journal of Medical Genetics, 2016, 59, 540-545.	0.7	21
153	A novel thymidine phosphorylase mutation in a Spanish MNGIE patient. Journal of the Neurological Sciences, 2005, 228, 35-39.	0.3	20
154	A polymorphic polymerase. Brain, 2006, 129, 1637-1639.	3.7	20
155	A novel tRNAVal mitochondrial DNA mutation causing MELAS. Journal of the Neurological Sciences, 2008, 270, 23-27.	0.3	20
156	Growth Differentiation Factor 15 is a potential biomarker of therapeutic response for TK2 deficient myopathy. Scientific Reports, 2020, 10, 10111.	1.6	20
157	Survival transcriptome in the coenzyme Q₁₀deficiency syndrome is acquired by epigenetic modifications: a modelling study for human coenzyme Q₁₀deficiencies. BMJ Open, 2013, 3, e002524.	0.8	19
158	Deoxynucleoside stress exacerbates the phenotype of a mouse model of mitochondrial neurogastrointestinal encephalopathy. Brain, 2014, 137, 1337-1349.	3.7	19
159	A Novel Mutation in PNPLA2 Leading to Neutral Lipid Storage Disease With Myopathy. Archives of Neurology, 2012, 69, 1190.	4.9	18
160	Cerebral Mitochondrial Microangiopathy Leads to Leukoencephalopathy in Mitochondrial Neurogastrointestinal Encephalopathy. American Journal of Neuroradiology, 2018, 39, 427-434.	1.2	18
161	Fatigue in primary genetic mitochondrial disease: No rest for the weary. Neuromuscular Disorders, 2019, 29, 895-902.	0.3	18
162	Leberâ€™s Hereditary Optic Neuropathy Mitochondrial DNA Mutations at Nucleotides 11778 and 3460 in Multiple Sclerosis. Ophthalmologica, 1999, 213, 171-175.	1.0	17

#	Article	IF	Citations
163	Analysis of mtDNA deletions in muscle by in situ hybridization. Muscle and Nerve, 2000, 23, 80-85.	1.0	17
164	Selective muscle fiber loss and molecular compensation in mitochondrial myopathy due to TK2 deficiency. Journal of the Neurological Sciences, 2008, 267, 137-141.	0.3	17
165	Thymidine Kinase 2 Deficiency-Induced Mitochondrial DNA Depletion Causes Abnormal Development of Adipose Tissues and Adipokine Levels in Mice. PLoS ONE, 2011, 6, e29691.	1.1	17
166	Attitudes toward prevention of mtDNA-related diseases through oocyte mitochondrial replacement therapy. Human Reproduction, 2016, 31, 1058-1065.	0.4	17
167	Bioavailability and cytosolic kinases modulate response to deoxynucleoside therapy in TK2 deficiency. EBioMedicine, 2019, 46, 356-367.	2.7	17
168	Leber hereditary optic neuropathy plus dystonia, and transverse myelitis due to double mutations in MT-ND4 and MT-ND6. Journal of Neurology, 2020, 267, 823-829.	1.8	17
169	Cytochrome oxidase deficiency: progress and problems. , 1994, , 91-115.		16
170	Paracrine Stimulation of Endothelial Cell Motility and Angiogenesis by Platelet-Derived Deoxyribose-1-Phosphate. Arteriosclerosis, Thrombosis, and Vascular Biology, 2010, 30, 2631-2638.	1.1	16
171	Cardiac transplantation in Friedreich Ataxia: Extended follow-up. Journal of the Neurological Sciences, 2017, 375, 471-473.	0.3	16
172	Alpha-1-Antitrypsin Promoter Improves the Efficacy of an Adeno-Associated Virus Vector for the Treatment of Mitochondrial Neurogastrointestinal Encephalomyopathy. Human Gene Therapy, 2019, 30, 985-998.	1.4	16
173	Disentangling (Epi)Genetic and Environmental Contributions to the Mitochondrial 3243A>G Mutation Phenotype. JAMA Neurology, 2016, 73, 923.	4.5	15
174	A Novel SUCLA2 Mutation Presenting as a Complex Childhood Movement Disorder. Journal of Child Neurology, 2017, 32, 246-250.	0.7	15
175	Oculopharyngeal muscular dystrophy, other ocular myopathies, and progressive external ophthalmoplegia. Neuromuscular Disorders, 1997, 7, S15-S21.	0.3	14
176	Assessment of Thymidine Phosphorylase Function: Measurement of Plasma Thymidine (and) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 222 T	0.4	14
177	Synergistic Deoxynucleoside and Gene Therapies for Thymidine Kinase 2 Deficiency. Annals of Neurology, 2021, 90, 640-652.	2.8	14
178	Thymidine Phosphorylase Gene Mutations Cause Mitochondrial Neurogastrointestinal Encephalomyopathy (MNGIE). Internal Medicine, 2006, 45, 1103-1103.	0.3	12
179	Recurrent myoglobinuria in a sporadic patient with a novel mitochondrial DNA tRNAIle mutation. Journal of the Neurological Sciences, 2011, 303, 39-42.	0.3	12
180	Measurement of Mitochondrial dNTP Pools. Methods in Molecular Biology, 2012, 837, 135-148.	0.4	12

#	Article	IF	Citations
181	Characterization of the human homozygous R182W POLG2 mutation in mitochondrial DNA depletion syndrome. PLoS ONE, 2018, 13, e0203198.	1.1	11
182	Efficacy of adeno-associated virus gene therapy in a MNGIE murine model enhanced by chronic exposure to nucleosides. EBioMedicine, 2020, 62, 103133.	2.7	11
183	Mitochondrial neurogastrointestinal encephalomyopathy and thymidine metabolism: results and hypotheses. Mitochondrion, 2002, 2, 143-147.	1.6	10
184	Altered gene transcription profiles in fibroblasts harboring either TK2 or DGUOK mutations indicate compensatory mechanisms. Experimental Cell Research, 2009, 315, 1429-1438.	1.2	9
185	Autocrine amplification of integrin Î±IIbÎ²3 activation and platelet adhesive responses by deoxyribose-1-phosphate. Thrombosis and Haemostasis, 2013, 109, 1108-1119.	1.8	9
186	Cardiomyopathy and altered integrin-actin signaling in Fhl1 mutant female mice. Human Molecular Genetics, 2019, 28, 209-219.	1.4	9
187	Rod-sparing paraneoplastic retinopathy, opsoclonus, and peripheral neuropathy due to small cell lung carcinoma. Neuro-Ophthalmology, 1997, 17, 101-105.	0.4	8
188	Achalasia as the Harbinger of a Novel Mitochondrial Disorder in Childhood. Journal of Pediatric Gastroenterology and Nutrition, 2005, 40, 512-517.	0.9	8
189	The m.3244G>A mutation in mtDNA is another cause of progressive external ophthalmoplegia. Neuromuscular Disorders, 2009, 19, 297-299.	0.3	8
190	Targeted impairment of thymidine kinase 2 expression in cells induces mitochondrial DNA depletion and reveals molecular mechanisms of compensation of mitochondrial respiratory activity. Biochemical and Biophysical Research Communications, 2011, 407, 333-338.	1.0	8
191	Mitochondrial Cardioencephalomyopathy Due to a Novel SCO2 Mutation in a Brazilian Patient. JAMA Neurology, 2013, 70, 258.	4.5	8
192	Growth differentiation factor-15 as a biomarker of strength and recovery in survivors of acute respiratory failure. Thorax, 2019, 74, 1099-1101.	2.7	7
193	Editing the Mitochondrial Genome. New England Journal of Medicine, 2020, 383, 1489-1491.	13.9	7
194	Advances in Thymidine Kinase 2 Deficiency: Clinical Aspects, Translational Progress, and Emerging Therapies. Journal of Neuromuscular Diseases, 2022, 9, 225-235.	1.1	6
195	RRM1 variants cause a mitochondrial DNA maintenance disorder via impaired de novo nucleotide synthesis. Journal of Clinical Investigation, 2022, 132, .	3.9	6
196	MtDNA maintenance and stability genes: MNGIE and mtDNA depletion syndromes. Topics in Current Genetics, 2004, , 177-200.	0.7	5
197	Mutation in an mtDNA Protein-Coding Gene. Journal of Child Neurology, 2013, 28, 264-268.	0.7	5
198	Implications of mitochondrial DNA mutations in human induced pluripotent stem cells. Nature Reviews Genetics, 2022, 23, 69-70.	7.7	5

#	Article	IF	Citations
199	Leukocyte cytokine responses in adult patients with mitochondrial DNA defects. Journal of Molecular Medicine, 2022, 100, 963-971.	1.7	5
200	Chapter 3 Molecular Genetic Basis of the Mitochondrial Encephalomyopathies. Blue Books of Practical Neurology, 2002, , 69-113.	0.1	4
201	VMA21 Deficiency: A Case of Myocyte Indigestion. Cell, 2009, 137, 213-215.	13.5	4
202	Diagnosis of mitochondrial neurogastrointestinal encephalomyopathy: Proposal of a clinical algorithm. Digestive and Liver Disease, 2014, 46, 664-665.	0.4	4
203	A De Novo Mutation in MTND6 Causes Generalized Dystonia in 2 Unrelated Children. Child Neurology Open, 2016, 3, 2329048X1562793.	0.5	4
204	Mitochondrial Diseases: A Clinical and Molecular History. Pediatric Neurology, 2016, 63, 3-5.	1.0	4
205	Mitochondrial Neurogastrointestinal Encephalomyopathy Disease (MNGIE). , 2019, , 205-222.		4
206	The North American mitochondrial disease registry. , 2020, 4, 81-90.		4
207	Risk mitigation behaviors to prevent infection in the mitochondrial disease community during the COVID-19 pandemic. Molecular Genetics and Metabolism Reports, 2022, 30, 100837.	0.4	4
208	232nd ENMC International Workshop: Recommendations for treatment of mitochondrial DNA maintenance disorders. 16 â€“ 18 June 2017, Heemskerk, The Netherlands.. Neuromuscular Disorders, 2022, , .	0.3	4
209	Recalcitrant Vomiting, Disturbed Eye Movements, and Leukoencephalopathy. Gastroenterology, 2009, 137, 1581-1861.	0.6	3
210	Stroke-Like Episodes in Mitochondrial Encephalopathy, Lactic Acidosis, and Stroke-Like Episodes (MELAS). , 2013, , 107-125.		3
211	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.	1.2	3
212	Cerebellar Ataxia and Deficiency. , 2013, 1, 1004.		3
213	Vitamin status and mitochondrial DNA mutations in patients with incomplete recovery from an epidemic optic neuropathy in Cuba. Neuro-Ophthalmology, 1996, 16, 355-360.	0.4	2
214	Metabolic Myopathies. , 2007, , 947-956.		2
215	Drug Effects in Patients with Mitochondrial Diseases. , 0, , 311-324.		2
216	Metabolic myopathies. Advances in Neurology, 2002, 88, 217-34.	0.8	2

#	Article	IF	Citations
217	Does increased superoxide dismutase activity really cause muscular dystrophy?. Annals of Neurology, 1999, 46, 135-135.	2.8	1
218	A First Step in Viral Gene Therapy for Muscular Dystrophy. Current Neurology and Neuroscience Reports, 2010, 10, 71-72.	2.0	1
219	Regulatory environment for novel therapeutic development in mitochondrial diseases. Journal of Inherited Metabolic Disease, 2021, 44, 292-300.	1.7	1
220	Mitochondrial Disorders. , 2008, , 1785-1798.		1
221	Mitochondrial Myopathies. , 2014, , 1335-1353.		1
222	Whole Exome Sequencing detects PYGM variants in two adults with McArdle disease. Journal of Physical Education and Sports Management, 2022, , mcs.a006173.	0.5	1
223	Autosomal dominant hereditary spastic paraplegia with axonal sensory motor polyneuropathy maps to chromosome 21q 22.3. International Journal of Neuroscience, 2016, 126, 1-7.	0.8	0
224	Mitochondrial Neurogastrointestinal Encephalomyopathy (MNGIE). , 2016, , 199-206.		0
225	Therapies Approaches in Mitochondrial Diseases. , 2021, , 273-305.		0
226	Mitochondrial Disorders. Neurological Disease and Therapy, 2005, , 256-283.	0.0	0
227	Primary Cerebellar CoQ10 Deficiency. , 2016, , 293-297.		0
228	Stroke-Like Episodes in Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, and Stroke-Like Episodes (MELAS). , 2017, , 117-134.		0
229	Visual memory failure presages conversion to <scp>MELAS</scp> phenotype. Annals of Clinical and Translational Neurology, 2022, , .	1.7	0