Michio Hirano

List of Publications by Year
in descending order

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229 papers	19,655 citations	7096 78 h-index	12272 133 g-index
258 all docs	258 docs citations	258 times ranked	17167 citing authors

#	Article	IF	Citations
1	Implications of mitochondrial DNA mutations in human induced pluripotent stem cells. Nature Reviews Genetics, 2022, 23, 69-70.	16.3	5
2	Whole Exome Sequencing detects PYGM variants in two adults with McArdle disease. Journal of Physical Education and Sports Management, 2022, , mcs.a006173.	1.2	1
3	Advances in Thymidine Kinase 2 Deficiency: Clinical Aspects, Translational Progress, and Emerging Therapies. Journal of Neuromuscular Diseases, 2022, 9, 225-235.	2.6	6
4	Risk mitigation behaviors to prevent infection in the mitochondrial disease community during the COVID-19 pandemic. Molecular Genetics and Metabolism Reports, 2022, 30, 100837.	1.1	4
5	Visual memory failure presages conversion to <scp>MELAS</scp> phenotype. Annals of Clinical and Translational Neurology, 2022, , .	3.7	0
6	232nd ENMC International Workshop: Recommendations for treatment of mitochondrial DNA maintenance disorders. 16 â€“ 18 June 2017, Heemskerk, The Netherlands.. Neuromuscular Disorders, 2022, , .	0.6	4
7	RRM1 variants cause a mitochondrial DNA maintenance disorder via impaired de novo nucleotide synthesis. Journal of Clinical Investigation, 2022, 132, .	8.2	6
8	Leukocyte cytokine responses in adult patients with mitochondrial DNA defects. Journal of Molecular Medicine, 2022, 100, 963-971.	3.9	5
9	SETX (senataxin), the helicase mutated in AOA2 and ALS4, functions in autophagy regulation. Autophagy, 2021, 17, 1889-1906.	9.1	34
10	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.	3.6	47
11	Therapies Approaches in Mitochondrial Diseases. , 2021, , 273-305.		0
12	Regulatory environment for novel therapeutic development in mitochondrial diseases. Journal of Inherited Metabolic Disease, 2021, 44, 292-300.	3.6	1
13	Circulating markers of NADH-reductive stress correlate with mitochondrial disease severity. Journal of Clinical Investigation, 2021, 131, .	8.2	95
14	Synergistic Deoxynucleoside and Gene Therapies for Thymidine Kinase 2 Deficiency. Annals of Neurology, 2021, 90, 640-652.	5.3	14
15	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
16	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.	3.6	17
17	Editing the Mitochondrial Genome. New England Journal of Medicine, 2020, 383, 1489-1491.	27.0	7
18	Efficacy of adeno-associated virus gene therapy in a MNGIE murine model enhanced by chronic exposure to nucleosides. EBioMedicine, 2020, 62, 103133.	6.1	11

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19	<scp>GGPS1</scp> Mutations Cause Muscular Dystrophy/Hearing Loss/Ovarian Insufficiency Syndrome. Annals of Neurology, 2020, 88, 332-347.	5.3	22
20	Growth Differentiation Factor 15 is a potential biomarker of therapeutic response for TK2 deficient myopathy. Scientific Reports, 2020, 10, 10111.	3.3	20
21	Intracellular calcium leak as a therapeutic target for RYR1-related myopathies. Acta Neuropathologica, 2020, 139, 1089-1104.	7.7	32
22	Successful liver transplantation in mitochondrial neurogastrointestinal encephalomyopathy (MNGIE). Molecular Genetics and Metabolism, 2020, 130, 58-64.	1.1	26
23	Mitochondrial diseases in North America. Neurology: Genetics, 2020, 6, e402.	1.9	38
24	Metabolic shift underlies recovery in reversible infantile respiratory chain deficiency. EMBO Journal, 2020, 39, e105364.	7.8	26
25	The North American mitochondrial disease registry. , 2020, 4, 81-90.		4
26	Bioavailability and cytosolic kinases modulate response to deoxynucleoside therapy in TK2 deficiency. EBioMedicine, 2019, 46, 356-367.	6.1	17
27	Dystonia-Ataxia with early handwriting deterioration in COQ8A mutation carriers: A case series and literature review. Parkinsonism and Related Disorders, 2019, 68, 8-16.	2.2	25
28	Fatigue in primary genetic mitochondrial disease: No rest for the weary. Neuromuscular Disorders, 2019, 29, 895-902.	0.6	18
29	Growth differentiation factor-15 as a biomarker of strength and recovery in survivors of acute respiratory failure. Thorax, 2019, 74, 1099-1101.	5.6	7
30	Deoxynucleoside Therapy for Thymidine Kinase 2â€“Deficient Myopathy. Annals of Neurology, 2019, 86, 293-303.	5.3	72
31	Human aging DNA methylation signatures are conserved but accelerated in cultured fibroblasts. Epigenetics, 2019, 14, 961-976.	2.7	36
32	Late-onset thymidine kinase 2 deficiency: a review of 18 cases. Orphanet Journal of Rare Diseases, 2019, 14, 100.	2.7	29
33	The clinical spectrum of the congenital myasthenic syndrome resulting from COL13A1 mutations. Brain, 2019, 142, 1547-1560.	7.6	30
34	Mitochondrial Neurogastrointestinal Encephalomyopathy Disease (MNGIE). , 2019, , 205-222.		4
35	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.	2.7	16
36	Advances in primary mitochondrial myopathies. Current Opinion in Neurology, 2019, 32, 715-721.	3.6	32

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37	Cardiomyopathy and altered integrin-actin signaling in Fhl1 mutant female mice. Human Molecular Genetics, 2019, 28, 209-219.	2.9	9
38	Cerebral Mitochondrial Microangiopathy Leads to Leukoencephalopathy in Mitochondrial Neurogastrointestinal Encephalopathy. American Journal of Neuroradiology, 2018, 39, 427-434.	2.4	18
39	Long-Term Sustained Effect of Liver-Targeted Adeno-Associated Virus Gene Therapy for Mitochondrial Neurogastrointestinal Encephalomyopathy. Human Gene Therapy, 2018, 29, 708-718.	2.7	39
40	Retrospective natural history of thymidine kinase 2 deficiency. Journal of Medical Genetics, 2018, 55, 515-521.	3.2	73
41	Diagnostic odyssey of patients with mitochondrial disease. Neurology: Genetics, 2018, 4, e230.	1.9	92
42	Three-Dimensional Analysis of Mitochondrial Crista Ultrastructure in a Patient with Leigh Syndrome by In Situ Cryoelectron Tomography. IScience, 2018, 6, 83-91.	4.1	60
43	Characterization of the human homozygous R182W POLG2 mutation in mitochondrial DNA depletion syndrome. PLoS ONE, 2018, 13, e0203198.	2.5	11
44	Emerging therapies for mitochondrial diseases. Essays in Biochemistry, 2018, 62, 467-481.	4.7	113
45	A novel complex neurological phenotype due to a homozygous mutation in FDX2. Brain, 2018, 141, 2289-2298.	7.6	29
46	USMG5 Ashkenazi Jewish founder mutation impairs mitochondrial complex V dimerization and ATP synthesis. Human Molecular Genetics, 2018, 27, 3305-3312.	2.9	45
47	Cardiac transplantation in Friedreich Ataxia: Extended follow-up. Journal of the Neurological Sciences, 2017, 375, 471-473.	0.6	16
48	Deoxycytidine and Deoxythymidine Treatment for Thymidine Kinase 2 Deficiency. Annals of Neurology, 2017, 81, 641-652.	5.3	89
49	Low-dose rapamycin extends lifespan in a mouse model of mtDNA depletion syndrome. Human Molecular Genetics, 2017, 26, 4588-4605.	2.9	70
50	International Workshop:. Neuromuscular Disorders, 2017, 27, 1126-1137.	0.6	58
51	A Novel SUCLA2 Mutation Presenting as a Complex Childhood Movement Disorder. Journal of Child Neurology, 2017, 32, 246-250.	1.4	15
52	Inhibition of NADPH oxidase 2 (NOX2) prevents sepsis-induced cardiomyopathy by improving calcium handling and mitochondrial function. JCI Insight, 2017, 2, .	5.0	83
53	Stroke-Like Episodes in Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, and Stroke-Like Episodes (MELAS). , 2017, , 117-134.		0
54	Autosomal dominant hereditary spastic paraplegia with axonal sensory motor polyneuropathy maps to chromosome 21q 22.3. International Journal of Neuroscience, 2016, 126, 1-7.	1.6	0

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55	Mitochondrial Neurogastrointestinal Encephalomyopathy (MNGIE). , 2016, , 199-206.		0
56	Inhibition of NAPDH Oxidase 2 (NOX2) Prevents Oxidative Stress and Mitochondrial Abnormalities Caused by Saturated Fat in Cardiomyocytes. PLoS ONE, 2016, 11, e0145750.	2.5	78
57	A De Novo Mutation in MTND6 Causes Generalized Dystonia in 2 Unrelated Children. Child Neurology Open, 2016, 3, 2329048X1562793.	1.1	4
58	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.	6.9	84
59	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.	7.8	71
60	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.	2.7	26
61	Genetic Drift Can Compromise Mitochondrial Replacement by Nuclear Transfer in Human Oocytes. Cell Stem Cell, 2016, 18, 749-754.	11.1	170
62	Nutritional interventions in primary mitochondrial disorders: Developing an evidence base. Molecular Genetics and Metabolism, 2016, 119, 187-206.	1.1	41
63	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.	1.3	21
64	Mitochondrial disease patients' perception of dietary supplements' use. Molecular Genetics and Metabolism, 2016, 119, 100-108.	1.1	24
65	TheCOQ2genotype predicts the severity of coenzyme Q₁₀deficiency. Human Molecular Genetics, 2016, 25, 4256-4265.	2.9	53
66	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.	6.2	146
67	Mitochondrial diseases. Nature Reviews Disease Primers, 2016, 2, 16080.	30.5	1,001
68	Disentangling (Epi)Genetic and Environmental Contributions to the Mitochondrial 3243A>G Mutation Phenotype. JAMA Neurology, 2016, 73, 923.	9.0	15
69	Mitochondrial Diseases: A Clinical and Molecular History. Pediatric Neurology, 2016, 63, 3-5.	2.1	4
70	Attitudes toward prevention of mtDNA-related diseases through oocyte mitochondrial replacement therapy. Human Reproduction, 2016, 31, 1058-1065.	0.9	17
71	MPV17 Loss Causes Deoxynucleotide Insufficiency and Slow DNA Replication in Mitochondria. PLoS Genetics, 2016, 12, e1005779.	3.5	67
72	Primary Cerebellar CoQ10 Deficiency. , 2016, , 293-297.		0

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73	Diagnosis and management of mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. Genetics in Medicine, 2015, 17, 689-701.	2.4	414
74	Allogeneic haematopoietic stem cell transplantation for mitochondrial neurogastrointestinal encephalomyopathy. Brain, 2015, 138, 2847-2858.	7.6	128
75	Deoxynucleoside stress exacerbates the phenotype of a mouse model of mitochondrial neurogastrointestinal encephalopathy. Brain, 2014, 137, 1337-1349.	7.6	19
76	Pathomechanisms in Coenzyme Q₁₀-Deficient Human Fibroblasts. Molecular Syndromology, 2014, 5, 163-169.	0.8	23
77	Clinical Presentations of Coenzyme Q10 Deficiency Syndrome. Molecular Syndromology, 2014, 5, 141-146.	0.8	38
78	Deoxypyrimidine monophosphate bypass therapy for thymidine kinase 2 deficiency. EMBO Molecular Medicine, 2014, 6, 1016-1027.	6.9	79
79	Branching Enzyme Deficiency. JAMA Neurology, 2014, 71, 41.	9.0	43
80	Thymidine Phosphorylase Participates in Platelet Signaling and Promotes Thrombosis. Circulation Research, 2014, 115, 997-1006.	4.5	37
81	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.	8.2	55
82	Administration of deoxyribonucleosides or inhibition of their catabolism as a pharmacological approach for mitochondrial DNA depletion syndrome. Human Molecular Genetics, 2014, 23, 2459-2467.	2.9	67
83	Diagnosis of mitochondrial neurogastrointestinal encephalomyopathy: Proposal of a clinical algorithm. Digestive and Liver Disease, 2014, 46, 664-665.	0.9	4
84	Mitochondrial Myopathies. , 2014, , 1335-1353.		1
85	Tissueâ€specific oxidative stress and loss of mitochondria in CoQâ€deficient Pdss2 mutant mice. FASEB Journal, 2013, 27, 612-621.	0.5	61
86	The clinical maze of mitochondrial neurology. Nature Reviews Neurology, 2013, 9, 429-444.	10.1	293
87	Nuclear genome transfer in human oocytes eliminates mitochondrial DNA variants. Nature, 2013, 493, 632-637.	27.8	223
88	New treatments for mitochondrial diseaseâ€”no time to drop our standards. Nature Reviews Neurology, 2013, 9, 474-481.	10.1	157
89	Survival transcriptome in the coenzyme Q₁₀deficiency syndrome is acquired by epigenetic modifications: a modelling study for human coenzyme Q₁₀deficiencies. BMJ Open, 2013, 3, e002524.	1.9	19
90	Mitochondrial Cardioencephalomyopathy Due to a Novel SCO2 Mutation in a Brazilian Patient. JAMA Neurology, 2013, 70, 258.	9.0	8

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91	Mutation in an mtDNA Protein-Coding Gene. Journal of Child Neurology, 2013, 28, 264-268.	1.4	5
92	TK2 mutation presenting as indolent myopathy. Neurology, 2013, 80, 504-506.	1.1	28
93	Autocrine amplification of integrin Î±IIbÎ²3 activation and platelet adhesive responses by deoxyribose-1-phosphate. Thrombosis and Haemostasis, 2013, 109, 1108-1119.	3.4	9
94	Stroke-Like Episodes in Mitochondrial Encephalopathy, Lactic Acidosis, and Stroke-Like Episodes (MELAS). , 2013, , 107-125.		3
95	Cerebellar Ataxia and Deficiency. , 2013, 1, 1004.		3
96	Haploinsufficiency ofCOQ4causes coenzyme Q₁₀deficiency. Journal of Medical Genetics, 2012, 49, 187-191.	3.2	95
97	Heterogeneity of Coenzyme Q₁₀Deficiency. Archives of Neurology, 2012, 69, 978-83.	4.5	192
98	A Novel Mutation in PNPLA2 Leading to Neutral Lipid Storage Disease With Myopathy. Archives of Neurology, 2012, 69, 1190.	4.5	18
99	Human mitochondrial DNA: roles of inherited and somatic mutations. Nature Reviews Genetics, 2012, 13, 878-890.	16.3	620
100	CoQ10 deficiencies and MNGIE: Two treatable mitochondrial disorders. Biochimica Et Biophysica Acta - General Subjects, 2012, 1820, 625-631.	2.4	83
101	Assessment of Thymidine Phosphorylase Function: Measurement of Plasma Thymidine (and) Tj ETQq1 1 0.784314 rgBT /Overlock 10 Tf	6.9	14
102	MPV17 Mutations Causing Adult-Onset Multisystemic Disorder With Multiple Mitochondrial DNA Deletions. Archives of Neurology, 2012, 69, 1648.	4.5	68
103	Measurement of Mitochondrial dNTP Pools. Methods in Molecular Biology, 2012, 837, 135-148.	0.9	12
104	Effects of Inhibiting CoQ10 Biosynthesis with 4-nitrobenzoate in Human Fibroblasts. PLoS ONE, 2012, 7, e30606.	2.5	40
105	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.	2.3	21
106	Recurrent myoglobinuria in a sporadic patient with a novel mitochondrial DNA tRNAIle mutation. Journal of the Neurological Sciences, 2011, 303, 39-42.	0.6	12
107	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.	2.1	8
108	Thymidine Kinase 2 Deficiency-Induced Mitochondrial DNA Depletion Causes Abnormal Development of Adipose Tissues and Adipokine Levels in Mice. PLoS ONE, 2011, 6, e29691.	2.5	17

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109	FGF-21 as a biomarker for muscle-manifesting mitochondrial respiratory chain deficiencies: a diagnostic study. Lancet Neurology, The, 2011, 10, 806-818.	10.2	352
110	Primary and secondary CoQ₁₀ deficiencies in humans. BioFactors, 2011, 37, 361-365.	5.4	96
111	MERRF and Kearnsâ€“Sayre overlap syndrome due to the mitochondrial DNA m.3291T>C mutation. Muscle and Nerve, 2011, 44, 448-451.	2.2	23
112	Senataxin mutations and amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2011, 12, 223-227.	2.1	81
113	Onset and organ specificity of Tk2 deficiency depends on Tk1 down-regulation and transcriptional compensation. Human Molecular Genetics, 2011, 20, 155-164.	2.9	30
114	Clinical and genetic spectrum of mitochondrial neurogastrointestinal encephalomyopathy. Brain, 2011, 134, 3326-3332.	7.6	191
115	A First Step in Viral Gene Therapy for Muscular Dystrophy. Current Neurology and Neuroscience Reports, 2010, 10, 71-72.	4.2	1
116	A Diagnostic Algorithm for Metabolic Myopathies. Current Neurology and Neuroscience Reports, 2010, 10, 118-126.	4.2	128
117	Coenzyme Q and mitochondrial disease. Developmental Disabilities Research Reviews, 2010, 16, 183-188.	2.9	157
118	Treatment of CoQ10 Deficient Fibroblasts with Ubiquinone, CoQ Analogs, and Vitamin C: Time- and Compound-Dependent Effects. PLoS ONE, 2010, 5, e11897.	2.5	92
119	Reactive oxygen species, oxidative stress, and cell death correlate with level of CoQ ₁₀ deficiency. FASEB Journal, 2010, 24, 3733-3743.	0.5	142
120	Paracrine Stimulation of Endothelial Cell Motility and Angiogenesis by Platelet-Derived Deoxyribose-1-Phosphate. Arteriosclerosis, Thrombosis, and Vascular Biology, 2010, 30, 2631-2638.	2.4	16
121	Therapeutic prospects for mitochondrial disease. Trends in Molecular Medicine, 2010, 16, 268-276.	6.7	97
122	Neutral lipid storage disease with subclinical myopathy due to a retrotransposal insertion in the PNPLA2 gene. Neuromuscular Disorders, 2010, 20, 397-402.	0.6	58
123	Protean Phenotypic Features of the A3243G Mitochondrial DNA Mutation. Archives of Neurology, 2009, 66, 85-91.	4.5	53
124	Molecular basis of infantile reversible cytochrome c oxidase deficiency myopathy. Brain, 2009, 132, 3165-3174.	7.6	112
125	Unbalanced deoxynucleotide pools cause mitochondrial DNA instability in thymidine phosphorylase-deficient mice. Human Molecular Genetics, 2009, 18, 714-722.	2.9	123
126	Altered gene transcription profiles in fibroblasts harboring either TK2 or DGUOK mutations indicate compensatory mechanisms. Experimental Cell Research, 2009, 315, 1429-1438.	2.6	9

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127	Clinical and genetic analysis of lipid storage myopathies. Muscle and Nerve, 2009, 39, 333-342.	2.2	74
128	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.	6.2	206
129	VMA21 Deficiency: A Case of Myocyte Indigestion. Cell, 2009, 137, 213-215.	28.9	4
130	ETFDH mutations, CoQ10 levels, and respiratory chain activities in patients with riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency. Neuromuscular Disorders, 2009, 19, 212-216.	0.6	118
131	The m.3244G>A mutation in mtDNA is another cause of progressive external ophthalmoplegia. Neuromuscular Disorders, 2009, 19, 297-299.	0.6	8
132	Recalcitrant Vomiting, Disturbed Eye Movements, and Leukoencephalopathy. Gastroenterology, 2009, 137, 1581-1861.	1.3	3
133	Pathogenesis and Treatment of Mitochondrial Disorders. Advances in Experimental Medicine and Biology, 2009, 652, 139-170.	1.6	31
134	Coenzyme Q10 Deficiencies in Neuromuscular Diseases. Advances in Experimental Medicine and Biology, 2009, 652, 117-128.	1.6	21
135	Human CoQ₁₀ deficiencies. BioFactors, 2008, 32, 113-118.	5.4	110
136	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.	6.2	290
137	Selective muscle fiber loss and molecular compensation in mitochondrial myopathy due to TK2 deficiency. Journal of the Neurological Sciences, 2008, 267, 137-141.	0.6	17
138	A novel tRNAVal mitochondrial DNA mutation causing MELAS. Journal of the Neurological Sciences, 2008, 270, 23-27.	0.6	20
139	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.	6.2	108
140	Mitochondrial DNA depletion syndrome due to mutations in the RRM2B gene. Neuromuscular Disorders, 2008, 18, 453-459.	0.6	87
141	Functional characterization of human COQ4, a gene required for Coenzyme Q10 biosynthesis. Biochemical and Biophysical Research Communications, 2008, 372, 35-39.	2.1	49
142	Gastrointestinal Dysmotility in Mitochondrial Neurogastrointestinal Encephalomyopathy Is Caused by Mitochondrial DNA Depletion. American Journal of Pathology, 2008, 173, 1120-1128.	3.8	100
143	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.	2.9	101
144	Respiratory chain dysfunction and oxidative stress correlate with severity of primary CoQ ₁₀ deficiency. FASEB Journal, 2008, 22, 1874-1885.	0.5	150

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145	A functionally dominant mitochondrial DNA mutation. Human Molecular Genetics, 2008, 17, 1814-1820.	2.9	104
146	Amyotrophic Lateral Sclerosis With Ragged-Red Fibers. Archives of Neurology, 2008, 65, 403-6.	4.5	28
147	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.	4.5	113
148	Mitochondrial Disorders. , 2008, , 1785-1798.		1
149	Missense mutation of the COQ2 gene causes defects of bioenergetics and de novo pyrimidine synthesis. Human Molecular Genetics, 2007, 16, 1091-1097.	2.9	129
150	A novel ECGF1 mutation in a Thai patient with mitochondrial neurogastrointestinal encephalomyopathy (MNGIE). Clinical Neurology and Neurosurgery, 2007, 109, 613-616.	1.4	21
151	The myopathic form of coenzyme Q10 deficiency is caused by mutations in the electron-transferring-flavoprotein dehydrogenase (ETFDH) gene. Brain, 2007, 130, 2037-2044.	7.6	298
152	Metabolic Myopathies. , 2007, , 947-956.		2
153	Thymidine and deoxyuridine accumulate in tissues of patients with mitochondrial neurogastrointestinal encephalomyopathy (MNGIE). FEBS Letters, 2007, 581, 3410-3414.	2.8	64
154	Human Coenzyme Q10 Deficiency. Neurochemical Research, 2007, 32, 723-727.	3.3	163
155	Mutations in coenzyme Q10 biosynthetic genes. Journal of Clinical Investigation, 2007, 117, 587-589.	8.2	89
156	A Mutation in Para-Hydroxybenzoate-Polyprenyl Transferase (COQ2) Causes Primary Coenzyme Q10 Deficiency. American Journal of Human Genetics, 2006, 78, 345-349.	6.2	322
157	Navajo Neurohepatopathy Is Caused by a Mutation in the MPV17 Gene. American Journal of Human Genetics, 2006, 79, 544-548.	6.2	158
158	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.	6.2	359
159	Mitochondrial Neurogastrointestinal Encephalomyopathy: Evidence of Mitochondrial DNA Depletion in the Small Intestine. Gastroenterology, 2006, 130, 893-901.	1.3	63
160	Human mitochondrial pyrophosphatase: cDNA cloning and analysis of the gene in patients with mtDNA depletion syndromes. Genomics, 2006, 87, 410-416.	2.9	22
161	Thymidine Phosphorylase Gene Mutations Cause Mitochondrial Neurogastrointestinal Encephalomyopathy (MNGIE). Internal Medicine, 2006, 45, 1103-1103.	0.7	12
162	Approaches to the treatment of mitochondrial diseases. Muscle and Nerve, 2006, 34, 265-283.	2.2	130

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163	A polymorphic polymerase. Brain, 2006, 129, 1637-1639.	7.6	20
164	Mitochondrial DNA Depletion and Thymidine Phosphate Pool Dynamics in a Cellular Model of Mitochondrial Neurogastrointestinal Encephalomyopathy. Journal of Biological Chemistry, 2006, 281, 22720-22728.	3.4	68
165	Autophagic Vacuoles with Sarcolemmal Features Delineate Danon Disease and Related Myopathies. Journal of Neuropathology and Experimental Neurology, 2005, 64, 513-522.	1.7	81
166	Achalasia as the Harbinger of a Novel Mitochondrial Disorder in Childhood. Journal of Pediatric Gastroenterology and Nutrition, 2005, 40, 512-517.	1.8	8
167	POLG mutations and Alpers syndrome. Annals of Neurology, 2005, 57, 921-923.	5.3	131
168	Late-onset MNGIE due to partial loss of thymidine phosphorylase activity. Annals of Neurology, 2005, 58, 649-652.	5.3	64
169	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.	7.1	273
170	Does Linezolid Cause Lactic Acidosis by Inhibiting Mitochondrial Protein Synthesis?. Clinical Infectious Diseases, 2005, 40, e113-e116.	5.8	123
171	Thymidine phosphorylase mutations cause instability of mitochondrial DNA. Gene, 2005, 354, 152-156.	2.2	49
172	Novel cell lines derived from adult human ventricular cardiomyocytes. Journal of Molecular and Cellular Cardiology, 2005, 39, 133-147.	1.9	236
173	Mitochondrial encephalomyopathies: an update. Neuromuscular Disorders, 2005, 15, 276-286.	0.6	130
174	A novel thymidine phosphorylase mutation in a Spanish MNGIE patient. Journal of the Neurological Sciences, 2005, 228, 35-39.	0.6	20
175	Mitochondrial Disorders. Neurological Disease and Therapy, 2005, , 256-283.	0.0	0
176	MRI of Five Patients with Mitochondrial Neurogastrointestinal Encephalomyopathy. American Journal of Roentgenology, 2004, 182, 1537-1541.	2.2	37
177	Definitive Diagnosis of Mitochondrial Neurogastrointestinal Encephalomyopathy by Biochemical Assays. Clinical Chemistry, 2004, 50, 120-124.	3.2	107
178	Mitochondrial Neurogastrointestinal Encephalomyopathy (MNGIE): A Disease of Two Genomes. Neurologist, 2004, 10, 8-17.	0.7	156
179	MtDNA maintenance and stability genes: MNGIE and mtDNA depletion syndromes. Topics in Current Genetics, 2004, , 177-200.	0.7	5
180	Primary coenzyme Q₁₀ deficiency and the brain. BioFactors, 2003, 18, 145-152.	5.4	49

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181	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.	2.8	161
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