Agnes Rotig

List of Publications by Year
in descending order

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		8159	11581
234	20,795	76	135
papers	citations	h-index	g-index

247	247	247	15931
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Biochemical and molecular investigations in respiratory chain deficiencies. Clinica Chimica Acta, 1994, 228, 35-51.	0.5	1,160
2	Aconitase and mitochondrial ironâ€“sulphur protein deficiency in Friedreich ataxia. Nature Genetics, 1997, 17, 215-217.	9.4	1,027
3	Mutation of a nuclear succinate dehydrogenase gene results in mitochondrial respiratory chain deficiency. Nature Genetics, 1995, 11, 144-149.	9.4	684
4	Persistent mitochondrial dysfunction and perinatal exposure to antiretroviral nucleoside analogues. Lancet, The, 1999, 354, 1084-1089.	6.3	627
5	Mutation of RRM2B, encoding p53-controlled ribonucleotide reductase (p53R2), causes severe mitochondrial DNA depletion. Nature Genetics, 2007, 39, 776-780.	9.4	478
6	Pearson's marrow-pancreas syndrome. A multisystem mitochondrial disorder in infancy.. Journal of Clinical Investigation, 1990, 86, 1601-1608.	3.9	417
7	Selective iron chelation in Friedreich ataxia: biologic and clinical implications. Blood, 2007, 110, 401-408.	0.6	407
8	Mitochondrial double-stranded RNA triggers antiviral signalling in humans. Nature, 2018, 560, 238-242.	13.7	397
9	MPV17 encodes an inner mitochondrial membrane protein and is mutated in infantile hepatic mitochondrial DNA depletion. Nature Genetics, 2006, 38, 570-575.	9.4	380
10	Effect of idebenone on cardiomyopathy in Friedreich's ataxia: a preliminary study. Lancet, The, 1999, 354, 477-479.	6.3	352
11	Quinone-responsive multiple respiratory-chain dysfunction due to widespread coenzyme Q10 deficiency. Lancet, The, 2000, 356, 391-395.	6.3	349
12	COQ6 mutations in human patients produce nephrotic syndrome with sensorineural deafness. Journal of Clinical Investigation, 2011, 121, 2013-2024.	3.9	343
13	The R22X Mutation of the SDHD Gene in Hereditary Paraganglioma Abolishes the Enzymatic Activity of Complex II in the Mitochondrial Respiratory Chain and Activates the Hypoxia Pathway. American Journal of Human Genetics, 2001, 69, 1186-1197.	2.6	339
14	Mutations of theSCO1Gene in Mitochondrial CytochromecOxidase Deficiency with Neonatalâ€Onset Hepatic Failure and Encephalopathy. American Journal of Human Genetics, 2000, 67, 1104-1109.	2.6	322
15	A mutant mitochondrial respiratory chain assembly protein causes complex III deficiency in patients with tubulopathy, encephalopathy and liver failure. Nature Genetics, 2001, 29, 57-60.	9.4	297
16	CABC1 Gene Mutations Cause Ubiquinone Deficiency with Cerebellar Ataxia and Seizures. American Journal of Human Genetics, 2008, 82, 623-630.	2.6	284
17	A mutation in the human heme A:farnesyltransferase gene (COX10) causes cytochrome c oxidase deficiency. Human Molecular Genetics, 2000, 9, 1245-1249.	1.4	261
18	Mutation of the fumarase gene in two siblings with progressive encephalopathy and fumarase deficiency.. Journal of Clinical Investigation, 1994, 93, 2514-2518.	3.9	254

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19	Large-Scale Deletion and Point Mutations of the Nuclear NDUFV1 and NDUFS1 Genes in Mitochondrial Complex I Deficiency. American Journal of Human Genetics, 2001, 68, 1344-1352.	2.6	243
20	Prenyldiphosphate synthase, subunit 1 (PDSS1) and OH-benzoate polyprenyltransferase (COQ2) mutations in ubiquinone deficiency and oxidative phosphorylation disorders. Journal of Clinical Investigation, 2007, 117, 765-772.	3.9	227
21	Functional Consequences of aSDHBGene Mutation in an Apparently Sporadic Pheochromocytoma. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 4771-4774.	1.8	210
22	Succinate dehydrogenase and human diseases: new insights into a well-known enzyme. European Journal of Human Genetics, 2002, 10, 289-291.	1.4	208
23	OPA1 links human mitochondrial genome maintenance to mtDNA replication and distribution. Genome Research, 2011, 21, 12-20.	2.4	207
24	Acute Infantile Liver Failure Due to Mutations in the TRMU Gene. American Journal of Human Genetics, 2009, 85, 401-407.	2.6	205
25	Spectrum of mitochondrial DNA rearrangements in the Pearson marrow-pancreas syndrome. Human Molecular Genetics, 1995, 4, 1327-1330.	1.4	193
26	Disabled early recruitment of antioxidant defenses in Friedreich's ataxia. Human Molecular Genetics, 2001, 10, 2061-2067.	1.4	176
27	Cytochrome oxidase in health and disease. Gene, 2002, 286, 53-63.	1.0	175
28	Mutant NDUFS3 subunit of mitochondrial complex I causes Leigh syndrome. Journal of Medical Genetics, 2004, 41, 14-17.	1.5	167
29	Mutations of the SCO1 Gene in Mitochondrial Cytochrome c Oxidase Deficiency with Neonatal-Onset Hepatic Failure and Encephalopathy. American Journal of Human Genetics, 2000, 67, 1104-1109.	2.6	157
30	Twinkle helicase (PEO1) gene mutation causes mitochondrial DNA depletion. Annals of Neurology, 2007, 62, 579-587.	2.8	157
31	Mutant NDUFV2 subunit of mitochondrial complex I causes early onset hypertrophic cardiomyopathy and encephalopathy. Human Mutation, 2003, 21, 582-586.	1.1	152
32	Epileptic phenotypes in children with respiratory chain disorders. Epilepsia, 2010, 51, 1225-1235.	2.6	152
33	Assessment of the mitochondrial respiratory chain. Lancet, The, 1991, 338, 60.	6.3	151
34	Coenzyme Q10 and idebenone in the therapy of respiratory chain diseases: rationale and comparative benefits. Molecular Genetics and Metabolism, 2002, 77, 21-30.	0.5	150
35	Deletion of mitochondrial DNA in a case of early-onset diabetes mellitus, optic atrophy, and deafness (Wolfram syndrome, MIM 222300).. Journal of Clinical Investigation, 1993, 91, 1095-1098.	3.9	149
36	Hepatic mitochondrial DNA deletion in alcoholics: Association with microvesicular steatosis. Gastroenterology, 1995, 108, 193-200.	0.6	144

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37	Mitochondrial DNA Depletion is a Prevalent Cause of Multiple Respiratory Chain Deficiency in Childhood. Journal of Pediatrics, 2007, 150, 531-534.e6.	0.9	143
38	Clinical presentations and laboratory investigations in respiratory chain deficiency. European Journal of Pediatrics, 1996, 155, 262-274.	1.3	136
39	Clinical presentation of mitochondrial disorders in childhood. Journal of Inherited Metabolic Disease, 1996, 19, 521-527.	1.7	135
40	The mitochondrial DNA G13513A MELAS mutation in the NADH dehydrogenase 5 gene is a frequent cause of Leigh-like syndrome with isolated complex I deficiency. Journal of Medical Genetics, 2003, 40, 188-191.	1.5	135
41	Clinical aspects of mitochondrial disorders. Journal of Inherited Metabolic Disease, 1992, 15, 448-455.	1.7	131
42	Antenatal manifestations of mitochondrial respiratory chain deficiency. Journal of Pediatrics, 2003, 143, 208-212.	0.9	129
43	Activation of Peroxisome Proliferator-Activated Receptor Pathway Stimulates the Mitochondrial Respiratory Chain and Can Correct Deficiencies in Patientsâ€™ Cells Lacking Its Components. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 1433-1441.	1.8	128
44	Human diseases with impaired mitochondrial protein synthesis. Biochimica Et Biophysica Acta - Bioenergetics, 2011, 1807, 1198-1205.	0.5	127
45	Superoxide-induced massive apoptosis in cultured skin fibroblasts harboring the neurogenic ataxia retinitis pigmentosa (NARP) mutation in the ATPase-6 gene of the mitochondrial DNA. Human Molecular Genetics, 2001, 10, 1221-1228.	1.4	126
46	Exome sequencing identifies MRPL3 mutation in mitochondrial cardiomyopathy. Human Mutation, 2011, 32, 1225-1231.	1.1	125
47	Mutations in GTPBP3 Cause a Mitochondrial Translation Defect Associated with Hypertrophic Cardiomyopathy, Lactic Acidosis, and Encephalopathy. American Journal of Human Genetics, 2014, 95, 708-720.	2.6	123
48	Yeast as a system for modeling mitochondrial disease mechanisms and discovering therapies. DMM Disease Models and Mechanisms, 2015, 8, 509-526.	1.2	115
49	Reference charts for respiratory chain activities in human tissues. Clinica Chimica Acta, 1994, 228, 53-70.	0.5	114
50	Riboflavin-responsive oxidative phosphorylation complex I deficiency caused by defective ACAD9: new function for an old gene. Brain, 2011, 134, 210-219.	3.7	113
51	Recurrent de novo mitochondrial DNA mutations in respiratory chain deficiency. Journal of Medical Genetics, 2003, 40, 896-899.	1.5	110
52	The Spectrum of Systemic Involvement in Adults Presenting with Renal Lesion and Mitochondrial tRNA(Leu) Gene Mutation. Journal of the American Society of Nephrology: JASN, 2003, 14, 2099-2108.	3.0	109
53	Site-specific deletions of the mitochondrial genome in the Pearson marrow-pancreas syndrome. Genomics, 1991, 10, 502-504.	1.3	108
54	Inborn errors of complex II â€“ Unusual human mitochondrial diseases. Biochimica Et Biophysica Acta - Bioenergetics, 2002, 1553, 117-122.	0.5	108

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55	Refractory epilepsy and mitochondrial dysfunction due to GM3 synthase deficiency. European Journal of Human Genetics, 2013, 21, 528-534.	1.4	107
56	Mitochondrial DNA rearrangements with onset as chronic diarrhea with villous atrophy. Journal of Pediatrics, 1994, 124, 63-70.	0.9	103
57	Individuals with mutations in XPNPEP3, which encodes a mitochondrial protein, develop a nephronophthisis-like nephropathy. Journal of Clinical Investigation, 2010, 120, 791-802.	3.9	102
58	Biochemical parameters for the diagnosis of mitochondrial respiratory chain deficiency in humans, and their lack of age-related changes. Biochemical Journal, 1998, 329, 249-254.	1.7	98
59	Absence of Relationship between the Level of Electron Transport Chain Activities and Aging in Human Skeletal Muscle. Biochemical and Biophysical Research Communications, 1996, 229, 536-539.	1.0	97
60	Coamplification of Nuclear Pseudogenes and Assessment of Heteroplasmy of Mitochondrial DNA Mutations. Biochemical and Biophysical Research Communications, 1998, 247, 57-59.	1.0	96
61	A mitochondrial cytochrome b mutation but no mutations of nuclearly encoded subunits in ubiquinol cytochrome c reductase (complex III) deficiency. Human Genetics, 1999, 104, 460-466.	1.8	95
62	Prevalence of rare mitochondrial DNA mutations in mitochondrial disorders. Journal of Medical Genetics, 2013, 50, 704-714.	1.5	95
63	A novel recurrent mitochondrial DNA mutation inND3 gene is associated with isolated complex I deficiency causing Leigh syndrome and dystonia. American Journal of Medical Genetics, Part A, 2007, 143A, 33-41.	0.7	94
64	Neonatal and delayed-onset liver involvement in disorders of oxidative phosphorylation. Journal of Pediatrics, 1997, 130, 817-822.	0.9	91
65	A common pattern of brain MRI imaging in mitochondrial diseases with complex I deficiency. Journal of Medical Genetics, 2011, 48, 16-23.	1.5	89
66	Recessive Mutations in TRMT10C Cause Defects in Mitochondrial RNA Processing and Multiple Respiratory Chain Deficiencies. American Journal of Human Genetics, 2016, 98, 993-1000.	2.6	89
67	Hair and Skin Disorders as Signs of Mitochondrial Disease. Pediatrics, 1999, 103, 428-433.	1.0	88
68	The kidney in mitochondrial cytopathies. Kidney International, 1997, 51, 1000-1007.	2.6	87
69	Endomyocardial biopsies for early detection of mitochondrial disorders in hypertrophic cardiomyopathies. Journal of Pediatrics, 1994, 124, 224-228.	0.9	86
70	TMEM126A, Encoding a Mitochondrial Protein, Is Mutated in Autosomal-Recessive Nonsyndromic Optic Atrophy. American Journal of Human Genetics, 2009, 84, 493-498.	2.6	85
71	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38.	3.6	85
72	Mutations in C12orf62, a Factor that Couples COX I Synthesis with Cytochrome c Oxidase Assembly, Cause Fatal Neonatal Lactic Acidosis. American Journal of Human Genetics, 2012, 90, 142-151.	2.6	84

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73	Biallelic Mutations in MRPS34 Lead to Instability of the Small Mitoribosomal Subunit and Leigh Syndrome. American Journal of Human Genetics, 2017, 101, 239-254.	2.6	83
74	Heart Hypertrophy and Function Are Improved by Idebenone in Friedreich's Ataxia. Free Radical Research, 2002, 36, 467-469.	1.5	81
75	Mutation in PNPT1 , which Encodes a Polyribonucleotide Nucleotidyltransferase, Impairs RNA Import into Mitochondria and Causes Respiratory-Chain Deficiency. American Journal of Human Genetics, 2012, 91, 912-918.	2.6	81
76	Deletion of the mitochondrial DNA in a case of de Toni-Debrï¿½-Fanconi syndrome and Pearson syndrome. Pediatric Nephrology, 1994, 8, 164-168.	0.9	80
77	Mutations in the tricarboxylic acid cycle enzyme, aconitase 2, cause either isolated or syndromic optic neuropathy with encephalopathy and cerebellar atrophy. Journal of Medical Genetics, 2014, 51, 834-838.	1.5	80
78	Fate and expression of the deleted mitochondrial DNA differ between human heteroplasmic skin fibroblast and Epstein-Barr virus-transformed lymphocyte cultures. Journal of Biological Chemistry, 1993, 268, 19369-76.	1.6	80
79	Nonsense mutations in the COX1 subunit impair the stability of respiratory chain complexes rather than their assembly. EMBO Journal, 2012, 31, 1293-1307.	3.5	79
80	Mutation screening of 75 candidate genes in 152 complex I deficiency cases identifies pathogenic variants in 16 genes includingNDUFB9. Journal of Medical Genetics, 2012, 49, 83-89.	1.5	78
81	A High Rate (20%â€“30%) of Parental Consanguinity in Cytochrome-Oxidase Deficiency. American Journal of Human Genetics, 1998, 63, 428-435.	2.6	77
82	Sengers syndrome: six novel AGK mutations in seven new families and review of the phenotypic and mutational spectrum of 29 patients. Orphanet Journal of Rare Diseases, 2014, 9, 119.	1.2	77
83	A novel mutation in the dihydrolipoamide dehydrogenase E3 subunit gene (DLD) resulting in an atypical form of Î±-ketoglutarate dehydrogenase deficiency. Human Mutation, 2005, 25, 323-324.	1.1	76
84	Mutations in mitochondrial ribosomal protein MRPL12 leads to growth retardation, neurological deterioration and mitochondrial translation deficiency. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2013, 1832, 1304-1312.	1.8	76
85	Genetic causes of mitochondrial DNA depletion in humans. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2009, 1792, 1103-1108.	1.8	74
86	Mutation dependance of the mitochondrial DNA copy number in the first stages of human embryogenesis. Human Molecular Genetics, 2013, 22, 1867-1872.	1.4	72
87	Genetic Features of Mitochondrial Respiratory Chain Disorders. Journal of the American Society of Nephrology: JASN, 2003, 14, 2995-3007.	3.0	71
88	Impaired Transferrin Receptor Palmitoylation and Recycling in Neurodegeneration with Brain Iron Accumulation. American Journal of Human Genetics, 2018, 102, 266-277.	2.6	69
89	Cytochrome c oxidase assay in minute amounts of human skeletal muscle using single wavelength spectrophotometers. Journal of Neuroscience Methods, 1998, 80, 107-111.	1.3	67
90	The neurogenic weakness, ataxia and retinitis pigmentosa (NARP) syndrome mtDNA mutation (T8993G) triggers muscle ATPase deficiency and hypocitrullinaemia. European Journal of Pediatrics, 1999, 158, 55-58.	1.3	64

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91	The measurement of the rotenone-sensitive NADH cytochrome c reductase activity in mitochondria isolated from minute amount of human skeletal muscle. Biochemical and Biophysical Research Communications, 1990, 173, 26-33.	1.0	63
92	Phenotypic variability in ARCA2 and identification of a core ataxic phenotype with slow progression. Orphanet Journal of Rare Diseases, 2013, 8, 173.	1.2	63
93	Mutations in MDH2, Encoding a Krebs Cycle Enzyme, Cause Early-Onset Severe Encephalopathy. American Journal of Human Genetics, 2017, 100, 151-159.	2.6	63
94	Isolation and characterization of mitochondria from human B lymphoblastoid cell lines. Biochemical and Biophysical Research Communications, 1992, 186, 16-23.	1.0	61
95	Bi-allelic Mutations in the Mitochondrial Ribosomal Protein MRPS2 Cause Sensorineural Hearing Loss, Hypoglycemia, and Multiple OXPHOS Complex Deficiencies. American Journal of Human Genetics, 2018, 102, 685-695.	2.6	61
96	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. Orphanet Journal of Rare Diseases, 2018, 13, 120.	1.2	61
97	Hepatic failure in disorders of oxidative phosphorylation with neonatal onset. Journal of Pediatrics, 1991, 119, 951-954.	0.9	60
98	Renal involvement in mitochondrial cytopathies. Pediatric Nephrology, 1996, 10, 368-373.	0.9	60
99	Infantile and pediatric quinone deficiency diseases. Mitochondrion, 2007, 7, S112-S121.	1.6	60
100	Beneficial effects of resveratrol on respiratory chain defects in patients' fibroblasts involve estrogen receptor and estrogen-related receptor alpha signaling. Human Molecular Genetics, 2014, 23, 2106-2119.	1.4	57
101	Molecular insights into Friedreich's ataxia and antioxidant-based therapies. Trends in Molecular Medicine, 2002, 8, 221-224.	3.5	56
102	Assay of mitochondrial respiratory chain complex I in human lymphocytes and cultured skin fibroblasts. Biochemical and Biophysical Research Communications, 2003, 301, 222-224.	1.0	56
103	Genotyping microsatellite DNA markers at putative disease loci in inbred/multiplex families with respiratory chain complexÂI deficiency allows rapid identification of a novel nonsense mutation (IVS1nt â^1) in the NDUFS4 gene in Leigh syndrome. Human Genetics, 2003, 112, 563-566.	1.8	54
104	NDUFS4 mutations cause Leigh syndrome with predominant brainstem involvement. Molecular Genetics and Metabolism, 2009, 97, 185-189.	0.5	54
105	DELETION OF BLOOD MITOCHONDRIAL DNA IN PANCYTOPENIA. Lancet, The, 1988, 332, 567-568.	6.3	53
106	Refractory anaemia and mitochondrial cytopathy in childhood. British Journal of Haematology, 1994, 87, 381-385.	1.2	53
107	Complex I assembly function and fatty acid oxidation enzyme activity of ACAD9 both contribute to disease severity in ACAD9 deficiency. Human Molecular Genetics, 2015, 24, 3238-3247.	1.4	53
108	Deletion of mitochondrial DNA in patient with chronic tubulointerstitial nephritis. Journal of Pediatrics, 1995, 126, 597-601.	0.9	52

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109	Mutations in Complex I Assembly Factor TMEM126B Result in Muscle Weakness and Isolated Complex I Deficiency. American Journal of Human Genetics, 2016, 99, 208-216.	2.6	51
110	Fluxes of Nicotinamide Adenine Dinucleotides through Mitochondrial Membranes in Human Cultured Cells. Journal of Biological Chemistry, 1996, 271, 14785-14790.	1.6	50
111	Pearson Syndrome in the Neonatal Period. Journal of Pediatric Hematology/Oncology, 2009, 31, 947-951.	0.3	50
112	An improved spectrophotometric assay of pyruvate dehydrogenase in lactate dehydrogenase contaminated mitochondrial preparations from human skeletal muscle. Clinica Chimica Acta, 1995, 240, 129-136.	0.5	49
113	Mitochondrial respiratory chain complex assembly and function during human fetal development. Molecular Genetics and Metabolism, 2008, 94, 120-126.	0.5	49
114	Phenotypic variability and identification of novel YARS2 mutations in YARS2 mitochondrial myopathy, lactic acidosis and sideroblastic anaemia. Orphanet Journal of Rare Diseases, 2013, 8, 193.	1.2	49
115	A constant and similar assembly defect of mitochondrial respiratory chain complex I allows rapid identification of NDUFS4 mutations in patients with Leigh syndrome. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 1062-1069.	1.8	46
116	Prenatal diagnosis of myopathy, encephalopathy, lactic acidosis, and stroke-like syndrome: contribution to understanding mitochondrial DNA segregation during human embryofetal development. Journal of Medical Genetics, 2006, 43, 788-792.	1.5	45
117	Respiratory chain deficiency presenting as congenital nephrotic syndrome. Pediatric Nephrology, 2005, 20, 465-469.	0.9	44
118	Compound heterozygosity for severe and hypomorphicNDUFS2mutations cause non-syndromic LHON-like optic neuropathy. Journal of Medical Genetics, 2017, 54, 346-356.	1.5	43
119	Mitochondria Transfection by Oligonucleotides Containing a Signal Peptide and Vectorized by Cationic Liposomes. Oligonucleotides, 2001, 11, 175-180.	4.4	42
120	Molecular analysis of ANT1, TWINKLE and POLG in patients with multiple deletions or depletion of mitochondrial DNA by a dHPLC-based assay. European Journal of Human Genetics, 2006, 14, 917-922.	1.4	42
121	Cerebral white matter disease in children may be caused by mitochondrial respiratory chain deficiency. Journal of Pediatrics, 2000, 136, 209-214.	0.9	41
122	A novel mutation in the human complex I NDUFS7 subunit associated with Leigh syndrome. Molecular Genetics and Metabolism, 2007, 90, 379-382.	0.5	41
123	Collated mutations in mitochondrial DNA (mtDNA) depletion syndrome (excluding the mitochondrial) Tj ETQq1 1 0.784314 rgBT /Ove 1109-1112.	1.8	41
124	Calcium signalling-dependent mitochondrial dysfunction and bioenergetics regulation in respiratory chain Complex II deficiency. Cell Death and Differentiation, 2010, 17, 1855-1866.	5.0	41
125	Duplications of mitochondrial DNA in Kearns-Sayre syndrome. Muscle and Nerve, 1995, 18, S154-S158.	1.0	40
126	A novel mutation of the NDUFS7 gene leads to activation of a cryptic exon and impaired assembly of mitochondrial complex I in a patient with Leigh syndrome. Molecular Genetics and Metabolism, 2007, 92, 104-108.	0.5	40

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127	Brain imaging in mitochondrial respiratory chain deficiency: combination of brain MRI features as a useful tool for genotype/phenotype correlations. Journal of Medical Genetics, 2014, 51, 429-435.	1.5	40
128	NDUFB8 Mutations Cause Mitochondrial Complex I Deficiency in Individuals with Leigh-like Encephalomyopathy. American Journal of Human Genetics, 2018, 102, 460-467.	2.6	40
129	Mutations in SURF1 are not specifically associated with Leigh syndrome. Journal of Medical Genetics, 2001, 38, 109-113.	1.5	40
130	Renal disease and mitochondrial genetics. Journal of Nephrology, 2003, 16, 286-92.	0.9	40
131	A case of Pearson syndrome associated with multiple renal cysts. Pediatric Nephrology, 1996, 10, 637-638.	0.9	39
132	Efficiency of metabolic screening in childhood cardiomyopathies. European Heart Journal, 1998, 19, 790-793.	1.0	39
133	Modeling of Antigenomic Therapy of Mitochondrial Diseases by Mitochondrially Addressed RNA Targeting a Pathogenic Point Mutation in Mitochondrial DNA. Journal of Biological Chemistry, 2014, 289, 13323-13334.	1.6	39
134	Biallelic PPA2 Mutations Cause Sudden Unexpected Cardiac Arrest in Infancy. American Journal of Human Genetics, 2016, 99, 666-673.	2.6	39
135	Respiratory Chain Deficiency in Alpers Syndrome. Neuropediatrics, 2001, 32, 150-152.	0.3	38
136	Idebenone treatment in Friedreich patients: One-year-long randomized placebo-controlled trial. Neurology, 2004, 62, 524-525.	1.5	38
137	Uridine preserves the expression of respiratory enzyme deficiencies in cultured fibroblasts. European Journal of Pediatrics, 1993, 152, 270-270.	1.3	37
138	New SUCLG1 patients expanding the phenotypic spectrum of this rare cause of mild methylmalonic aciduria. Mitochondrion, 2010, 10, 335-341.	1.6	37
139	Molecular diagnostics of mitochondrial disorders. Biochimica Et Biophysica Acta - Bioenergetics, 2004, 1659, 129-135.	0.5	36
140	Bi-Allelic UQCRFS1 Variants Are Associated with Mitochondrial Complex III Deficiency, Cardiomyopathy, and Alopecia Totalis. American Journal of Human Genetics, 2020, 106, 102-111.	2.6	36
141	The investigation of respiratory chain disorders in heart using endomyocardial biopsies. Journal of Inherited Metabolic Disease, 1993, 16, 541-544.	1.7	35
142	Deoxyguanosine kinase mutations and combined deficiencies of the mitochondrial respiratory chain in patients with hepatic involvement. Molecular Genetics and Metabolism, 2005, 86, 462-465.	0.5	34
143	1H MRS spectroscopy evidence of cerebellar high lactate in mitochondrial respiratory chain deficiency. Molecular Genetics and Metabolism, 2008, 93, 85-88.	0.5	34
144	Genetic bases of mitochondrial respiratory chain disorders. Diabetes and Metabolism, 2010, 36, 97-107.	1.4	34

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145	Poor Correlations in the Levels of Pathogenic Mitochondrial DNA Mutations in Polar Bodies versus Oocytes and Blastomeres in Humans. American Journal of Human Genetics, 2011, 88, 494-498.	2.6	34
146	Quinone analogs prevent enzymes targeted in Friedreich ataxia from ironâ€induced injury in vitro. BioFactors, 1999, 9, 247-251.	2.6	31
147	For debate: defective mitochondria, free radicals, cell death, aging-reality or myth-ochondria?. Mechanisms of Ageing and Development, 2000, 114, 201-206.	2.2	31
148	Tubulopathy and pancytopaenia with normal pancreatic function: A variant of Pearson syndrome. European Journal of Medical Genetics, 2009, 52, 23-26.	0.7	31
149	Mutation in the mitochondrial translation elongation factor EFTs results in severe infantile liver failure. Journal of Hepatology, 2012, 56, 294-297.	1.8	31
150	Macular dystrophy, diabetes, and deafness associated with a large mitochondrial dma deletion. American Journal of Ophthalmology, 1998, 125, 100-103.	1.7	30
151	Coenzyme Q 10 Depletion is Comparatively Less Detrimental to Human Cultured Skin Fibroblasts than Respiratory Chain Complex Deficiencies. Free Radical Research, 2002, 36, 375-379.	1.5	30
152	Loss of MTX2 causes mandibuloacral dysplasia and links mitochondrial dysfunction to altered nuclear morphology. Nature Communications, 2020, 11, 4589.	5.8	30
153	Cytochrome c oxidase deficiency presenting as recurrent neonatal myoglobinuria. Neuromuscular Disorders, 1995, 5, 285-289.	0.3	29
154	Biochemical, genetic and immunoblot analyses of 17 patients with an isolated cytochrome c oxidase deficiency. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 1999, 1455, 35-44.	1.8	29
155	Caenorhabditis elegans, a pluricellular model organism to screen new genes involved in mitochondrial genome maintenance. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2010, 1802, 765-773.	1.8	29
156	The first founder DGUOK mutation associated with hepatocerebral mitochondrial DNA depletion syndrome. Molecular Genetics and Metabolism, 2009, 97, 221-226.	0.5	28
157	Revisiting Pitfalls, Problems and Tentative Solutions for Assaying [General Articles] Mitochondrial Respiratory Chain Complex III in Human Samples. Current Medicinal Chemistry, 2004, 11, 233-239.	1.2	27
158	Massive and exclusive pontocerebellar damage in mitochondrial disease and NUBPL mutations. Neurology, 2012, 79, 391-391.	1.5	27
159	High incidence and variable clinical outcome of cardiac hypertrophy due to ACAD9 mutations in childhood. European Journal of Human Genetics, 2016, 24, 1112-1116.	1.4	27
160	Expression of respiratory chain deficiencies in human cultured cells. Neuromuscular Disorders, 1993, 3, 605-608.	0.3	25
161	Liver cytochrome c oxidase deficiency in a case of neonatal-onset hepatic failure. European Journal of Pediatrics, 1994, 153, 190-194.	1.3	25
162	Persistent hypocitrullinaemia as a marker for mtDNA NARP T 8993 G mutation?. Journal of Inherited Metabolic Disease, 1998, 21, 216-219.	1.7	24

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163	Haploinsufficiency of the GPD2 gene in a patient with nonsyndromic mental retardation. Human Genetics, 2009, 124, 649-658.	1.8	24
164	Mouse models for mitochondrial diseases. Human Molecular Genetics, 2016, 25, R115-R122.	1.4	24
165	New evidence of a mitochondrial genetic background paradox: Impact of the J haplogroup on the A3243G mutation. BMC Medical Genetics, 2008, 9, 41.	2.1	23
166	The human MSH5 (MutS Homolog 5) protein localizes to mitochondria and protects the mitochondrial genome from oxidative damage. Mitochondrion, 2012, 12, 654-665.	1.6	23
167	A multi-center comparison of diagnostic methods for the biochemical evaluation of suspected mitochondrial disorders. Mitochondrion, 2013, 13, 36-43.	1.6	23
168	Data from Artificial Models of Mitochondrial DNA Disorders Are Not Always Applicable to Humans. Cell Reports, 2014, 7, 933-934.	2.9	23
169	The Consequences of a Mild Respiratory Chain Deficiency on Substrate Competitive Oxidation in Human Mitochondria. Biochemical and Biophysical Research Communications, 1997, 236, 643-646.	1.0	22
170	Another observation with VATER association and a complex IV respiratory chain deficiency. European Journal of Medical Genetics, 2006, 49, 71-77.	0.7	21
171	High predictive value of brain MRI imaging in primary mitochondrial respiratory chain deficiency. Journal of Medical Genetics, 2018, 55, 378-383.	1.5	21
172	Mitochondrial Respiratory Chain Dysfunction Caused by Coenzyme Q Deficiency. Methods in Enzymology, 2004, 382, 81-88.	0.4	20
173	Toward genotype phenotype correlations in GFM1 mutations. Mitochondrion, 2012, 12, 242-247.	1.6	20
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