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	Heterogeneity of PNPT1 neuroimaging: mitochondriopathy, interferonopathy or both?. Journal of Medical Genetics, 2022, 59, 204-208.	1.5	6
2	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38.	3.6	85
3	Heterozygous <scp>PNPT1</scp> Variants Cause Spinocerebellar Ataxia Type 25. Annals of Neurology, 2022, 92, 122-137.	2.8	8
4	Neonatal gene therapy achieves sustained disease rescue of maple syrup urine disease in mice. Nature Communications, 2022, 13, .	5.8	8
5	A retrospective study on the efficacy of prenatal diagnosis for pregnancies at risk of mitochondrial DNA disorders. Genetics in Medicine, 2021, 23, 720-731.	1.1	5
6	Novel FARS2 variants in patients with early onset encephalopathy with or without epilepsy associated with long survival. European Journal of Human Genetics, 2021, 29, 533-538.	1.4	8
7	Cerebral blood flow and acute episodes of Leigh syndrome in neurometabolic disorders. Developmental Medicine and Child Neurology, 2021, 63, 705-711.	1.1	6
8	Novel NDUFA12 variants are associated with isolated complex I defect and variable clinical manifestation. Human Mutation, 2021, 42, 699-710.	1.1	12
9	Inherited glycosylphosphatidylinositol defects cause the rare Emm-negative blood phenotype and developmental disorders. Blood, 2021, 137, 3660-3669.	0.6	18
10	Haematological characteristics and spontaneous haematological recovery in Pearson syndrome. British Journal of Haematology, 2021, 193, 1283-1287.	1.2	8
11	Defective palmitoylation of transferrin receptor triggers iron overload in Friedreich ataxia fibroblasts. Blood, 2021, 137, 2090-2102.	0.6	16
12	Quantitative Susceptibility Mapping in Woodhouseâ€Sakati Syndrome. Annals of Neurology, 2021, 90, 324-325.	2.8	1
13	Biallelic mutations in the <i>SARS2</i> gene presenting as congenital sideroblastic anemia. Haematologica, 2021, 106, 3202-3205.	1.7	2
14	Variants in the MIPEP gene presenting with complex neurological phenotype without cardiomyopathy, impair OXPHOS protein maturation and lead to a reduced OXPHOS abundance in patient cells. Molecular Genetics and Metabolism, 2021, 134, 267-273.	0.5	4
15	Biallelic <i>IARS2</i> mutations presenting as sideroblastic anemia. Haematologica, 2021, 106, 0-0.	1.7	3
16	The natural history of infantile mitochondrial DNA depletion syndrome due to RRM2B deficiency. Genetics in Medicine, 2020, 22, 199-209.	1.1	14
17	Clinical, neuroimaging and biochemical findings in patients and patient fibroblasts expressing ten novel GFM1 mutations. Human Mutation, 2020, 41, 397-402.	1.1	10
18	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

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19	Improving post-natal detection of mitochondrial DNA mutations. Expert Review of Molecular Diagnostics, 2020, 20, 1003-1008.	1.5	2
20	Loss of MTX2 causes mandibuloacral dysplasia and links mitochondrial dysfunction to altered nuclear morphology. Nature Communications, 2020, 11, 4589.	5.8	30
21	Defects in Galactose Metabolism and Glycoconjugate Biosynthesis in a UDP-Glucose Pyrophosphorylase-Deficient Cell Line Are Reversed by Adding Galactose to the Growth Medium. International Journal of Molecular Sciences, 2020, 21, 2028.	1.8	7
22	Evidence of diaphragmatic dysfunction with severe alveolar hypoventilation syndrome in mitochondrial respiratory chain deficiency. Neuromuscular Disorders, 2020, 30, 593-598.	0.3	2
23	Mutations in the MRPS28 gene encoding the small mitoribosomal subunit protein bS1m in a patient with intrauterine growth retardation, craniofacial dysmorphism and multisystemic involvement. Human Molecular Genetics, 2019, 28, 1445-1462.	1.4	19
24	Expanding and Underscoring the Hepatoâ€Encephalopathic Phenotype of QIL1/MIC13. Hepatology, 2019, 70, 1066-1070.	3.6	17
25	Impaired Transferrin Receptor Palmitoylation and Recycling in Neurodegeneration with Brain Iron Accumulation. American Journal of Human Genetics, 2018, 102, 266-277.	2.6	69
26	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
27	High predictive value of brain MRI imaging in primary mitochondrial respiratory chain deficiency. Journal of Medical Genetics, 2018, 55, 378-383.	1.5	21
28	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
29	Pitfalls in molecular diagnosis of Friedreich ataxia. European Journal of Medical Genetics, 2018, 61, 455-458.	0.7	5
30	Segregation of mitochondrial DNA mutations in the human placenta: implication for prenatal diagnosis of mtDNA disorders. Journal of Medical Genetics, 2018, 55, 131-136.	1.5	11
31	Inhibition of mitochondrial translation in fibroblasts from a patient expressing the KARS p.(Pro228Leu) variant and presenting with sensorineural deafness, developmental delay, and lactic acidosis. Human Mutation, 2018, 39, 2047-2059.	1.1	14
32	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
33	Mitochondrial double-stranded RNA triggers antiviral signalling in humans. Nature, 2018, 560, 238-242.	13.7	397
34	Compound heterozygosity for severe and hypomorphicNDUFS2mutations cause non-syndromic LHON-like optic neuropathy. Journal of Medical Genetics, 2017, 54, 346-356.	1.5	43
35	Further delineation of a rare recessive encephalomyopathy linked to mutations in <scp>GFER</scp> thanks to data sharing of whole exome sequencing data. Clinical Genetics, 2017, 92, 188-198.	1.0	20
36	No correlation between mtDNA amount and methylation levels at the CpG island of POLG exon 2 in wild-type and mutant human differentiated cells. Journal of Medical Genetics, 2017, 54, 324-329.	1.5	5

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37	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
38	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
39	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
40	Combined use ofSaccharomyces cerevisiae,Caenorhabditis elegansand patient fibroblasts leads to the identification of clofilium tosylate as a potential therapeutic chemical against POLG-related diseases. Human Molecular Genetics, 2016, 25, 715-727.	1.4	18
41	Biallelic PPA2 Mutations Cause Sudden Unexpected Cardiac Arrest in Infancy. American Journal of Human Genetics, 2016, 99, 666-673.	2.6	39
42	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
43	Mouse models for mitochondrial diseases. Human Molecular Genetics, 2016, 25, R115-R122.	1.4	24
44	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
45	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
46	Unusual clinical expression and long survival of a pseudouridylate synthase (PUS1) mutation into adulthood. European Journal of Human Genetics, 2015, 23, 880-882.	1.4	20
47	Yeast as a system for modeling mitochondrial disease mechanisms and discovering therapies. DMM Disease Models and Mechanisms, 2015, 8, 509-526.	1.2	115
48	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
49	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
50	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
51	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
52	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
53	Genetics of mitochondrial respiratory chain deficiencies. Revue Neurologique, 2014, 170, 309-322.	0.6	8
54	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

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55	Data from Artificial Models of Mitochondrial DNA Disorders Are Not Always Applicable to Humans. Cell Reports, 2014, 7, 933-934.	2.9	23
56	Respiratory chain deficiencies. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1651-1666.	1.0	18
57	A multi-center comparison of diagnostic methods for the biochemical evaluation of suspected mitochondrial disorders. Mitochondrion, 2013, 13, 36-43.	1.6	23
58	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
59	Refractory epilepsy and mitochondrial dysfunction due to GM3 synthase deficiency. European Journal of Human Genetics, 2013, 21, 528-534.	1.4	107
60	A novel mutation in STXBP1 causing epileptic encephalopathy (late onset infantile spasms) with partial respiratory chain complex IV deficiency. European Journal of Medical Genetics, 2013, 56, 683-685.	0.7	11
61	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
62	Secondary Mitochondrial Respiratory Chain Defect Can Delay Accurate PFIC2 Diagnosis. JIMD Reports, 2013, 14, 17-21.	0.7	8
63	Prevalence of rare mitochondrial DNA mutations in mitochondrial disorders. Journal of Medical Genetics, 2013, 50, 704-714.	1.5	95
64	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
65	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
66	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
67	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
68	Massive and exclusive pontocerebellar damage in mitochondrial disease and NUBPL mutations. Neurology, 2012, 79, 391-391.	1.5	27
69	Maternal uniparental disomy of chromosome 2 in a patient with a DGUOK mutation associated with hepatocerebral mitochondrial DNA depletion syndrome. Molecular Genetics and Metabolism, 2012, 107, 700-704.	0.5	14
70	Mutation in the mitochondrial translation elongation factor EFTs results in severe infantile liver failure. Journal of Hepatology, 2012, 56, 294-297.	1.8	31
71	Toward genotype phenotype correlations in GFM1 mutations. Mitochondrion, 2012, 12, 242-247.	1.6	20
72	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

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73	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
74	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
75	Defects of the Respiratory Chain. , 2012, , 223-238.		7
76	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
77	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
78	Normal oxidative phosphorylation in intestinal smooth muscle of childhood chronic intestinal pseudo-obstruction. Neurogastroenterology and Motility, 2011, 23, 24-e1.	1.6	11
79	Human diseases with impaired mitochondrial protein synthesis. Biochimica Et Biophysica Acta - Bioenergetics, 2011, 1807, 1198-1205.	0.5	127
80	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
81	Exome sequencing identifies MRPL3 mutation in mitochondrial cardiomyopathy. Human Mutation, 2011, 32, 1225-1231.	1.1	125
82	OPA1 links human mitochondrial genome maintenance to mtDNA replication and distribution. Genome Research, 2011, 21, 12-20.	2.4	207
83	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
84	COQ6 mutations in human patients produce nephrotic syndrome with sensorineural deafness. Journal of Clinical Investigation, 2011, 121, 2013-2024.	3.9	343
85	News in Ubiquinone Biosynthesis. Chemistry and Biology, 2010, 17, 415-416.	6.2	3
86	Epileptic phenotypes in children with respiratory chain disorders. Epilepsia, 2010, 51, 1225-1235.	2.6	152
87	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
88	Mitochondrial ND5 mutations mimicking brainstem tectal glioma. Neurology, 2010, 75, 93-93.	1.5	8
89	Genetic bases of mitochondrial respiratory chain disorders. Diabetes and Metabolism, 2010, 36, 97-107.	1.4	34
90	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

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91	New SUCLG1 patients expanding the phenotypic spectrum of this rare cause of mild methylmalonic aciduria. Mitochondrion, 2010, 10, 335-341.	1.6	37
92	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
93	Haploinsufficiency of the GPD2 gene in a patient with nonsyndromic mental retardation. Human Genetics, 2009, 124, 649-658.	1.8	24
94	Multiple OXPHOS deficiency in the liver of a patient with CblA methylmalonic aciduria sensitive to vitamin B₁₂. Journal of Inherited Metabolic Disease, 2009, 32, 159-162.	1.7	13
95	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
96	Acute Infantile Liver Failure Due to Mutations in the TRMU Gene. American Journal of Human Genetics, 2009, 85, 401-407.	2.6	205
97	Tubulopathy and pancytopaenia with normal pancreatic function: A variant of Pearson syndrome. European Journal of Medical Genetics, 2009, 52, 23-26.	0.7	31
98	Genetic causes of mitochondrial DNA depletion in humans. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2009, 1792, 1103-1108.	1.8	74
99	Collated mutations in mitochondrial DNA (mtDNA) depletion syndrome (excluding the mitochondrial) Tj ETQq1 1 0.784314 rgBT /Ove 1109-1112.	1.8	41
100	NDUFS4 mutations cause Leigh syndrome with predominant brainstem involvement. Molecular Genetics and Metabolism, 2009, 97, 185-189.	0.5	54
101	The first founder DGUOK mutation associated with hepatocerebral mitochondrial DNA depletion syndrome. Molecular Genetics and Metabolism, 2009, 97, 221-226.	0.5	28
102	Pearson Syndrome in the Neonatal Period. Journal of Pediatric Hematology/Oncology, 2009, 31, 947-951.	0.3	50
103	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
104	CABC1 Gene Mutations Cause Ubiquinone Deficiency with Cerebellar Ataxia and Seizures. American Journal of Human Genetics, 2008, 82, 623-630.	2.6	284
105	1H MRS spectroscopy evidence of cerebellar high lactate in mitochondrial respiratory chain deficiency. Molecular Genetics and Metabolism, 2008, 93, 85-88.	0.5	34
106	Variable outcome of growth hormone administration in respiratory chain deficiency. Molecular Genetics and Metabolism, 2008, 93, 195-199.	0.5	10
107	Mitochondrial respiratory chain complex assembly and function during human fetal development. Molecular Genetics and Metabolism, 2008, 94, 120-126.	0.5	49
108	The regulation of PTC containing transcripts of the human NDUFS4 gene of complex I of respiratory chain and the impact of pathological mutations. Biochimie, 2008, 90, 1452-1460.	1.3	11

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109	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
110	Selective iron chelation in Friedreich ataxia: biologic and clinical implications. Blood, 2007, 110, 401-408.	0.6	407
111	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
112	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
113	Infantile and pediatric quinone deficiency diseases. Mitochondrion, 2007, 7, S112-S121.	1.6	60
114	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
115	Twinkle helicase (PEO1) gene mutation causes mitochondrial DNA depletion. Annals of Neurology, 2007, 62, 579-587.	2.8	157
116	Mutation of RRM2B, encoding p53-controlled ribonucleotide reductase (p53R2), causes severe mitochondrial DNA depletion. Nature Genetics, 2007, 39, 776-780.	9.4	478
117	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
118	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
119	Another observation with VATER association and a complex IV respiratory chain deficiency. European Journal of Medical Genetics, 2006, 49, 71-77.	0.7	21
120	Respiratory chain deficiency in a female with Aicardi-GoutiÃ¨res syndrome. Developmental Medicine and Child Neurology, 2006, 48, 227-230.	1.1	14
121	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
122	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
123	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
124	Mitochondrial nephrology. , 2006, , 197-207.		2
125	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
126	Respiratory chain deficiency presenting as congenital nephrotic syndrome. Pediatric Nephrology, 2005, 20, 465-469.	0.9	44

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127	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
128	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
129	Mutant NDUFS3 subunit of mitochondrial complex I causes Leigh syndrome. Journal of Medical Genetics, 2004, 41, 14-17.	1.5	167
130	Molecular diagnostics of mitochondrial disorders. Biochimica Et Biophysica Acta - Bioenergetics, 2004, 1659, 129-135.	0.5	36
131	Mitochondrial Respiratory Chain Dysfunction Caused by Coenzyme Q Deficiency. Methods in Enzymology, 2004, 382, 81-88.	0.4	20
132	Idebenone treatment in Friedreich patients: One-year-long randomized placebo-controlled trial. Neurology, 2004, 62, 524-525.	1.5	38
133	Mitochondrial Complex I Deficiency in Humans. Current Genomics, 2004, 5, 137-146.	0.7	7
134	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
135	Mutant NDUFV2 subunit of mitochondrial complex I causes early onset hypertrophic cardiomyopathy and encephalopathy. Human Mutation, 2003, 21, 582-586.	1.1	152
136	Mitochondrial activities in human cultured skin fibroblasts contaminated by Mycoplasma hyorhinis. BMC Biochemistry, 2003, 4, 15.	4.4	11
137	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
138	Antenatal manifestations of mitochondrial respiratory chain deficiency. Journal of Pediatrics, 2003, 143, 208-212.	0.9	129
139	Genetic Features of Mitochondrial Respiratory Chain Disorders. Journal of the American Society of Nephrology: JASN, 2003, 14, 2995-3007.	3.0	71
140	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
141	Recurrent de novo mitochondrial DNA mutations in respiratory chain deficiency. Journal of Medical Genetics, 2003, 40, 896-899.	1.5	110
142	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
143	Renal disease and mitochondrial genetics. Journal of Nephrology, 2003, 16, 286-92.	0.9	40
144	Cell complementation using Genebridge 4 human:rodent hybrids for physical mapping of novel mitochondrial respiratory chain deficiency genes. Human Molecular Genetics, 2002, 11, 3273-3281.	1.4	14

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145	Functional Consequences of aSDHBGene Mutation in an Apparently Sporadic Pheochromocytoma. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 4771-4774.	1.8	210
146	Expressed Sequence Tag Database Screening for Identification of Human Genes. Methods in Enzymology, 2002, 353, 566-574.	0.4	1
147	Heart Hypertrophy and Function Are Improved by Idebenone in Friedreich's Ataxia. Free Radical Research, 2002, 36, 467-469.	1.5	81
148	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
149	Inborn errors of complex II â€“ Unusual human mitochondrial diseases. Biochimica Et Biophysica Acta - Bioenergetics, 2002, 1553, 117-122.	0.5	108
150	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
151	Molecular insights into Friedreich's ataxia and antioxidant-based therapies. Trends in Molecular Medicine, 2002, 8, 221-224.	3.5	56
152	Cytochrome oxidase in health and disease. Gene, 2002, 286, 53-63.	1.0	175
153	Succinate dehydrogenase and human diseases: new insights into a well-known enzyme. European Journal of Human Genetics, 2002, 10, 289-291.	1.4	208
154	Expression Study of Genes Involved in Iron Metabolism in Human Tissues. Biochemical and Biophysical Research Communications, 2001, 281, 804-809.	1.0	2
155	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
156	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
157	Mitochondria Transfection by Oligonucleotides Containing a Signal Peptide and Vectorized by Cationic Liposomes. Oligonucleotides, 2001, 11, 175-180.	4.4	42
158	Prenatal diagnosis of respiratory chain deficiency by direct mutation screening. Prenatal Diagnosis, 2001, 21, 602-604.	1.1	15
159	Sequence variations in the NDUFA1 gene encoding a subunit of complex I of the respiratory chain. Journal of Inherited Metabolic Disease, 2001, 24, 15-27.	1.7	13
160	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
161	Human cultured skin fibroblasts survive profound inherited ubiquinone depletion. Free Radical Research, 2001, 35, 11-21.	1.5	17
162	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

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163	Disabled early recruitment of antioxidant defenses in Friedreich's ataxia. Human Molecular Genetics, 2001, 10, 2061-2067.	1.4	176
164	Respiratory Chain Deficiency in Alpers Syndrome. Neuropediatrics, 2001, 32, 150-152.	0.3	38
165	Mutations in SURF1 are not specifically associated with Leigh syndrome. Journal of Medical Genetics, 2001, 38, 109-113.	1.5	40
166	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
167	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
168	Screening Human EST Database for Identification of Candidate Genes in Respiratory Chain Deficiency. Molecular Genetics and Metabolism, 2000, 69, 223-232.	0.5	6
169	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
170	Quinone-responsive multiple respiratory-chain dysfunction due to widespread coenzyme Q10 deficiency. Lancet, The, 2000, 356, 391-395.	6.3	349
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