AgnÃ¨s RÃ¶tig

List of Publications by Year
in descending order

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194	16,052	13078	18028
papers	citations	h-index	g-index

205	205	205	15404
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Clinical, Neuroimaging, and Metabolic Footprint of the Neurodevelopmental Disorder Caused by Monoallelic HK1 Variants. Neurology: Genetics, 2024, 10, .	2.5	0
2	Systematic analysis of NDUFAF6 in complex I assembly and mitochondrial disease. Nature Metabolism, 2024, 6, 1128-1142.	11.4	2
3	Polyradiculoneuritis on MRI. Neurology, 2024, 102, .	1.1	0
4	Distinct Clinical Courses and Shortened Lifespans in Childhood-Onset DNA Polymerase Gamma Deficiency. Neurology: Genetics, 2024, 10, .	2.5	0
5	Preliminary Hydrogeological Investigations for Sustainable Development in the Courel Mountains UNESCO Global Geopark (NW Spain). Advances in Karst Science, 2023, , 249-254.	0.0	0
6	Novel ELAC2 Mutations in Individuals Presenting with Variably Severe Neurological Disease in the Presence or Absence of Cardiomyopathy. Life, 2023, 13, 445.	2.5	3
7	A shared pattern of altered gene expression in human embryos affected by mitochondrial diseases. Human Reproduction, 2023, 38, 992-1002.	0.9	2
8	Heterogeneity of PNPT1 neuroimaging: mitochondriopathy, interferonopathy or both?. Journal of Medical Genetics, 2022, 59, 204-208.	3.6	6
9	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38.	8.5	107
10	Heterozygous <scp>PNPT1</scp> Variants Cause Spinocerebellar Ataxia Type 25. Annals of Neurology, 2022, 92, 122-137.	5.8	10
11	Neonatal gene therapy achieves sustained disease rescue of maple syrup urine disease in mice. Nature Communications, 2022, 13, .	13.2	10
12	Fruit drop and bloom return according to time of metamitron application for chemical thinning in â€˜Red Chiefâ€™ apple. Acta Horticulturae, 2022, , 25-30.	0.2	0
13	A retrospective study on the efficacy of prenatal diagnosis for pregnancies at risk of mitochondrial DNA disorders. Genetics in Medicine, 2021, 23, 720-731.	2.4	7
14	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.	2.9	8
15	Cerebral blood flow and acute episodes of Leigh syndrome in neurometabolic disorders. Developmental Medicine and Child Neurology, 2021, 63, 705-711.	2.7	6
16	Inherited glycosylphosphatidylinositol defects cause the rare Emm-negative blood phenotype and developmental disorders. Blood, 2021, 137, 3660-3669.	1.4	20
17	Haematological characteristics and spontaneous haematological recovery in Pearson syndrome. British Journal of Haematology, 2021, 193, 1283-1287.	2.7	8
18	Defective palmitoylation of transferrin receptor triggers iron overload in Friedreich ataxia fibroblasts. Blood, 2021, 137, 2090-2102.	1.4	20

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19	Quantitative Susceptibility Mapping in Woodhouseâ€Sakati Syndrome. Annals of Neurology, 2021, 90, 324-325.	5.8	1
20	Biallelic mutations in the <i>SARS2</i> gene presenting as congenital sideroblastic anemia. Haematologica, 2021, 106, 3202-3205.	3.5	3
21	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.	2.2	5
22	Biallelic <i>IARS2</i> mutations presenting as sideroblastic anemia. Haematologica, 2021, 106, 0-0.	3.5	3
23	The natural history of infantile mitochondrial DNA depletion syndrome due to RRM2B deficiency. Genetics in Medicine, 2020, 22, 199-209.	2.4	17
24	Clinical, neuroimaging and biochemical findings in patients and patient fibroblasts expressing ten novel GFM1 mutations. Human Mutation, 2020, 41, 397-402.	2.8	13
25	Bi-Allelic UQCRFS1 Variants Are Associated with Mitochondrial Complex III Deficiency, Cardiomyopathy, and Alopecia Totalis. American Journal of Human Genetics, 2020, 106, 102-111.	6.1	39
26	Improving post-natal detection of mitochondrial DNA mutations. Expert Review of Molecular Diagnostics, 2020, 20, 1003-1008.	3.4	2
27	Loss of MTX2 causes mandibuloacral dysplasia and links mitochondrial dysfunction to altered nuclear morphology. Nature Communications, 2020, 11, 4589.	13.2	33
28	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.	4.2	8
29	Evidence of diaphragmatic dysfunction with severe alveolar hypoventilation syndrome in mitochondrial respiratory chain deficiency. Neuromuscular Disorders, 2020, 30, 593-598.	0.7	2
30	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.	3.0	20
31	Expanding and Underscoring the Hepatoâ€Encephalopathic Phenotype of QIL1/MIC13. Hepatology, 2019, 70, 1066-1070.	8.1	19
32	Impaired Transferrin Receptor Palmitoylation and Recycling in Neurodegeneration with Brain Iron Accumulation. American Journal of Human Genetics, 2018, 102, 266-277.	6.1	76
33	NDUFB8 Mutations Cause Mitochondrial Complex I Deficiency in Individuals with Leigh-like Encephalomyopathy. American Journal of Human Genetics, 2018, 102, 460-467.	6.1	42
34	High predictive value of brain MRI imaging in primary mitochondrial respiratory chain deficiency. Journal of Medical Genetics, 2018, 55, 378-383.	3.6	21
35	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.	6.1	64
36	Pitfalls in molecular diagnosis of Friedreich ataxia. European Journal of Medical Genetics, 2018, 61, 455-458.	1.3	7

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37	Segregation of mitochondrial DNA mutations in the human placenta: implication for prenatal diagnosis of mtDNA disorders. Journal of Medical Genetics, 2018, 55, 131-136.	3.6	11
38	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.	2.8	15
39	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. Orphanet Journal of Rare Diseases, 2018, 13, 120.	2.8	66
40	Mitochondrial double-stranded RNA triggers antiviral signalling in humans. Nature, 2018, 560, 238-242.	36.2	445
41	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.	3.6	5
42	Mutations in MDH2, Encoding a Krebs Cycle Enzyme, Cause Early-Onset Severe Encephalopathy. American Journal of Human Genetics, 2017, 100, 151-159.	6.1	69
43	Biallelic Mutations in MRPS34 Lead to Instability of the Small Mitoribosomal Subunit and Leigh Syndrome. American Journal of Human Genetics, 2017, 101, 239-254.	6.1	86
44	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.	6.1	52
45	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.	3.0	19
46	Biallelic PPA2 Mutations Cause Sudden Unexpected Cardiac Arrest in Infancy. American Journal of Human Genetics, 2016, 99, 666-673.	6.1	42
47	Recessive Mutations in TRMT10C Cause Defects in Mitochondrial RNA Processing and Multiple Respiratory Chain Deficiencies. American Journal of Human Genetics, 2016, 99, 246.	6.1	26
48	Recessive Mutations in TRMT10C Cause Defects in Mitochondrial RNA Processing and Multiple Respiratory Chain Deficiencies. American Journal of Human Genetics, 2016, 98, 993-1000.	6.1	92
49	Mouse models for mitochondrial diseases. Human Molecular Genetics, 2016, 25, R115-R122.	3.0	26
50	High incidence and variable clinical outcome of cardiac hypertrophy due to ACAD9 mutations in childhood. European Journal of Human Genetics, 2016, 24, 1112-1116.	2.9	29
51	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.	3.0	55
52	Unusual clinical expression and long survival of a pseudouridylate synthase (PUS1) mutation into adulthood. European Journal of Human Genetics, 2015, 23, 880-882.	2.9	22
53	Yeast as a system for modeling mitochondrial disease mechanisms and discovering therapies. DMM Disease Models and Mechanisms, 2015, 8, 509-526.	2.4	120
54	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.	2.8	79

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55	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.	3.6	83
56	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.	3.5	40
57	Data from Artificial Models of Mitochondrial DNA Disorders Are Not Always Applicable to Humans. Cell Reports, 2014, 7, 933-934.	6.3	23
58	Respiratory chain deficiencies. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1651-1666.	0.3	18
59	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.	2.8	50
60	Refractory epilepsy and mitochondrial dysfunction due to GM3 synthase deficiency. European Journal of Human Genetics, 2013, 21, 528-534.	2.9	110
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.	3.8	80
62	Secondary Mitochondrial Respiratory Chain Defect Can Delay Accurate PFIC2 Diagnosis. JIMD Reports, 2013, 14, 17-21.	1.2	8
63	Prevalence of rare mitochondrial DNA mutations in mitochondrial disorders. Journal of Medical Genetics, 2013, 50, 704-714.	3.6	105
64	Mutation dependance of the mitochondrial DNA copy number in the first stages of human embryogenesis. Human Molecular Genetics, 2013, 22, 1867-1872.	3.0	73
65	Phenotypic variability in ARCA2 and identification of a core ataxic phenotype with slow progression. Orphanet Journal of Rare Diseases, 2013, 8, 173.	2.8	65
66	Nonsense mutations in the COX1 subunit impair the stability of respiratory chain complexes rather than their assembly. EMBO Journal, 2012, 31, 1293-1307.	8.2	83
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.	3.6	79
68	Massive and exclusive pontocerebellar damage in mitochondrial disease and NUBPL mutations. Neurology, 2012, 79, 391-391.	1.1	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.	2.2	17
70	Mutation in the mitochondrial translation elongation factor EFTs results in severe infantile liver failure. Journal of Hepatology, 2012, 56, 294-297.	3.9	36
71	Toward genotype phenotype correlations in GFM1 mutations. Mitochondrion, 2012, 12, 242-247.	3.6	23
72	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.	6.1	85

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73	The human MSH5 (MutS Homolog 5) protein localizes to mitochondria and protects the mitochondrial genome from oxidative damage. Mitochondrion, 2012, 12, 654-665.	3.6	24
74	Defects of the Respiratory Chain. , 2012, , 223-238.		7
75	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.	6.1	89
76	Riboflavin-responsive oxidative phosphorylation complex I deficiency caused by defective ACAD9: new function for an old gene. Brain, 2011, 134, 210-219.	8.0	114
77	Human diseases with impaired mitochondrial protein synthesis. Biochimica Et Biophysica Acta - Bioenergetics, 2011, 1807, 1198-1205.	1.6	129
78	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.	6.1	34
79	Exome sequencing identifies MRPL3 mutation in mitochondrial cardiomyopathy. Human Mutation, 2011, 32, 1225-1231.	2.8	128
80	COQ6 mutations in human patients produce nephrotic syndrome with sensorineural deafness. Journal of Clinical Investigation, 2011, 121, 2013-2024.	8.2	347
81	News in Ubiquinone Biosynthesis. Chemistry and Biology, 2010, 17, 415-416.	6.2	3
82	Epileptic phenotypes in children with respiratory chain disorders. Epilepsia, 2010, 51, 1225-1235.	4.6	155
83	InstabilitÃ© du gÃ©nome mitochondrial et pathologies associÃ©es. Medecine/Sciences, 2010, 26, 171-176.	0.2	6
84	PrÃ©venir la transmission des mutations de lâ€™ADN mitochondrial : mythe ou rÃ©alitÃ© ?. Medecine/Sciences, 2010, 26, 897-899.	0.2	1
85	Genetic bases of mitochondrial respiratory chain disorders. Diabetes and Metabolism, 2010, 36, 97-107.	3.1	35
86	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.	3.8	30
87	New SUCLG1 patients expanding the phenotypic spectrum of this rare cause of mild methylmalonic aciduria. Mitochondrion, 2010, 10, 335-341.	3.6	38
88	Individuals with mutations in XPNPEP3, which encodes a mitochondrial protein, develop a nephronophthisis-like nephropathy. Journal of Clinical Investigation, 2010, 120, 791-802.	8.2	103
89	Haploinsufficiency of the GPD2 gene in a patient with nonsyndromic mental retardation. Human Genetics, 2009, 124, 649-658.	3.8	25
90	Acute Infantile Liver Failure Due to Mutations in the TRMU Gene. American Journal of Human Genetics, 2009, 85, 401-407.	6.1	210

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91	Tubulopathy and pancytopaenia with normal pancreatic function: A variant of Pearson syndrome. European Journal of Medical Genetics, 2009, 52, 23-26.	1.3	32
92	Genetic causes of mitochondrial DNA depletion in humans. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2009, 1792, 1103-1108.	3.8	76
93	NDUFS4 mutations cause Leigh syndrome with predominant brainstem involvement. Molecular Genetics and Metabolism, 2009, 97, 185-189.	2.2	55
94	Pearson Syndrome in the Neonatal Period. Journal of Pediatric Hematology/Oncology, 2009, 31, 947-951.	0.6	52
95	Cytopathies mitochondriales. Bulletin De L'Academie Nationale De Medecine, 2009, 193, 19-43.	0.1	1
96	New evidence of a mitochondrial genetic background paradox: Impact of the J haplogroup on the A3243G mutation. BMC Medical Genetics, 2008, 9, 41.	2.0	23
97	CABC1 Gene Mutations Cause Ubiquinone Deficiency with Cerebellar Ataxia and Seizures. American Journal of Human Genetics, 2008, 82, 623-630.	6.1	287
98	Mitochondrial respiratory chain complex assembly and function during human fetal development. Molecular Genetics and Metabolism, 2008, 94, 120-126.	2.2	50
99	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.	2.9	11
100	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.	3.6	129
101	Selective iron chelation in Friedreich ataxia: biologic and clinical implications. Blood, 2007, 110, 401-408.	1.4	412
102	A novel mutation in the human complex I NDUFS7 subunit associated with Leigh syndrome. Molecular Genetics and Metabolism, 2007, 90, 379-382.	2.2	41
103	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.	2.2	40
104	Infantile and pediatric quinone deficiency diseases. Mitochondrion, 2007, 7, S112-S121.	3.6	60
105	p53R2: rÃ©paration de lâ€™ADN ou synthÃ¨se de lâ€™ADN mitochondrialÂ?. Medecine/Sciences, 2007, 23, 803-805.	0.5	2
106	Twinkle helicase (PEO1) gene mutation causes mitochondrial DNA depletion. Annals of Neurology, 2007, 62, 579-587.	5.8	158
107	Mutation of RRM2B, encoding p53-controlled ribonucleotide reductase (p53R2), causes severe mitochondrial DNA depletion. Nature Genetics, 2007, 39, 776-780.	20.4	483
108	Mitochondrial DNA Depletion is a Prevalent Cause of Multiple Respiratory Chain Deficiency in Childhood. Journal of Pediatrics, 2007, 150, 531-534.e6.	2.2	147

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109	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.	8.2	230
110	Another observation with VATER association and a complex IV respiratory chain deficiency. European Journal of Medical Genetics, 2006, 49, 71-77.	1.3	22
111	MPV17 encodes an inner mitochondrial membrane protein and is mutated in infantile hepatic mitochondrial DNA depletion. Nature Genetics, 2006, 38, 570-575.	20.4	385
112	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.	2.9	42
113	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.	2.8	80
114	Homogeneous Catalysis. Understanding the Art. Von Piet W. N. M. van Leeuwen.. Angewandte Chemie, 2005, 117, 1321-1321.	2.1	0
115	Respiratory chain deficiency presenting as congenital nephrotic syndrome. Pediatric Nephrology, 2005, 20, 465-469.	1.8	44
116	Large Aperture Space Telescopes in Formation: Modeling, Metrology, and Control. Journal of the Astronautical Sciences, 2005, 53, 391-412.	1.4	11
117	Accumulation de mutations de lâ€™ADN mitochondrial chez la souris : un modÃ¨le idÃ©al de vieillissement. Medecine/Sciences, 2004, 20, 847-847.	0.2	0
118	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.	2.5	28
119	Molecular diagnostics of mitochondrial disorders. Biochimica Et Biophysica Acta - Bioenergetics, 2004, 1659, 129-135.	1.6	36
120	Mitochondrial Respiratory Chain Dysfunction Caused by Coenzyme Q Deficiency. Methods in Enzymology, 2004, 382, 81-88.	1.7	21
121	Cytopathies mitochondriales. EMC - Neurologie, 2004, 1, 1-10.	0.0	0
122	Idebenone treatment in Friedreich patients: One-year-long randomized placebo-controlled trial. Neurology, 2004, 62, 524-525.	1.1	40
123	Mitochondrial Complex I Deficiency in Humans. Current Genomics, 2004, 5, 137-146.	1.6	7
124	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.	3.8	54
125	Mutant NDUFV2 subunit of mitochondrial complex I causes early onset hypertrophic cardiomyopathy and encephalopathy. Human Mutation, 2003, 21, 582-586.	2.8	152
126	Mitochondrial activities in human cultured skin fibroblasts contaminated by Mycoplasma hyorhinis. BMC Biochemistry, 2003, 4, 15.	4.3	11

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127	Assay of mitochondrial respiratory chain complex I in human lymphocytes and cultured skin fibroblasts. Biochemical and Biophysical Research Communications, 2003, 301, 222-224.	2.2	56
128	Antenatal manifestations of mitochondrial respiratory chain deficiency. Journal of Pediatrics, 2003, 143, 208-212.	2.2	129
129	Genetic Features of Mitochondrial Respiratory Chain Disorders. Journal of the American Society of Nephrology: JASN, 2003, 14, 2995-3007.	0.5	75
130	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.	0.5	110
131	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.	3.0	14
132	Functional Consequences of aSDHBGene Mutation in an Apparently Sporadic Pheochromocytoma. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 4771-4774.	3.6	211
133	Expressed Sequence Tag Database Screening for Identification of Human Genes. Methods in Enzymology, 2002, 353, 566-574.	1.7	1
134	Heart Hypertrophy and Function Are Improved by Idebenone in Friedreich's Ataxia. Free Radical Research, 2002, 36, 467-469.	3.3	82
135	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.	3.3	30
136	Inborn errors of complex II â€“ Unusual human mitochondrial diseases. Biochimica Et Biophysica Acta - Bioenergetics, 2002, 1553, 117-122.	1.6	109
137	Coenzyme Q10 and idebenone in the therapy of respiratory chain diseases: rationale and comparative benefits. Molecular Genetics and Metabolism, 2002, 77, 21-30.	2.2	154
138	Molecular insights into Friedreich's ataxia and antioxidant-based therapies. Trends in Molecular Medicine, 2002, 8, 221-224.	7.1	56
139	Cytochrome oxidase in health and disease. Gene, 2002, 286, 53-63.	2.3	179
140	Succinate dehydrogenase and human diseases: new insights into a well-known enzyme. European Journal of Human Genetics, 2002, 10, 289-291.	2.9	216
141	Expression Study of Genes Involved in Iron Metabolism in Human Tissues. Biochemical and Biophysical Research Communications, 2001, 281, 804-809.	2.2	2
142	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.	6.1	248
143	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.	6.1	341
144	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.	20.4	300

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145	Human cultured skin fibroblasts survive profound inherited ubiquinone depletion. Free Radical Research, 2001, 35, 11-21.	3.3	17
146	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.	3.0	126
147	Respiratory Chain Deficiency in Alpers Syndrome. Neuropediatrics, 2001, 32, 150-152.	0.8	38
148	For debate: defective mitochondria, free radicals, cell death, aging-reality or myth-ochondria?. Mechanisms of Ageing and Development, 2000, 114, 201-206.	4.6	34
149	A mutation in the human heme A:farnesyltransferase gene (COX10) causes cytochrome c oxidase deficiency. Human Molecular Genetics, 2000, 9, 1245-1249.	3.0	264
150	Screening Human EST Database for Identification of Candidate Genes in Respiratory Chain Deficiency. Molecular Genetics and Metabolism, 2000, 69, 223-232.	2.2	6
151	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.	6.1	159
152	Cerebral white matter disease in children may be caused by mitochondrial respiratory chain deficiency. Journal of Pediatrics, 2000, 136, 209-214.	2.2	41
153	Respiratory Chain Deficiency Presenting as Recurrent Myoglobinuria in Childhood. Neuropediatrics, 1999, 30, 42-44.	0.8	13
154	Quinone analogs prevent enzymes targeted in Friedreich ataxia from ironâ€induced injury in vitro. BioFactors, 1999, 9, 247-251.	5.5	31
155	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.	3.8	95
156	Unusual presentation of Kearns-Sayre syndrome in early childhood. Pediatric Neurology, 1999, 21, 830-831.	2.1	13
157	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.	3.8	29
158	Effect of idebenone on cardiomyopathy in Friedreich's ataxia: a preliminary study. Lancet, The, 1999, 354, 477-479.	12.1	355
159	Persistent mitochondrial dysfunction and perinatal exposure to antiretroviral nucleoside analogues. Lancet, The, 1999, 354, 1084-1089.	12.1	628
160	Iron overload and mitochondrial diseases. Lancet, The, 1998, 351, 1286-1287.	12.1	17
161	A High Rate (20%â€“30%) of Parental Consanguinity in Cytochrome-Oxidase Deficiency. American Journal of Human Genetics, 1998, 63, 428-435.	6.1	78
162	Coamplification of Nuclear Pseudogenes and Assessment of Heteroplasmy of Mitochondrial DNA Mutations. Biochemical and Biophysical Research Communications, 1998, 247, 57-59.	2.2	96

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163	Macular dystrophy, diabetes, and deafness associated with a large mitochondrial dma deletion. American Journal of Ophthalmology, 1998, 125, 100-103.	3.4	30
164	The Consequences of a Mild Respiratory Chain Deficiency on Substrate Competitive Oxidation in Human Mitochondria. Biochemical and Biophysical Research Communications, 1997, 236, 643-646.	2.2	22
165	Sequence and structure of the human OXA1L gene and its upstream elements. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 1997, 1361, 6-10.	3.8	8
166	Neonatal and delayed-onset liver involvement in disorders of oxidative phosphorylation. Journal of Pediatrics, 1997, 130, 817-822.	2.2	94
167	Aconitase and mitochondrial ironâ€“sulphur protein deficiency in Friedreich ataxia. Nature Genetics, 1997, 17, 215-217.	20.4	1,035
168	The kidney in mitochondrial cytopathies. Kidney International, 1997, 51, 1000-1007.	5.4	87
169	No mitochondrial cytochrome oxidase (COX) gene mutations in 18 cases of COX deficiency. Human Genetics, 1997, 101, 247.	3.8	18
170	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.	2.2	98
171	A case of Pearson syndrome associated with multiple renal cysts. Pediatric Nephrology, 1996, 10, 637-638.	1.8	39
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