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	Aconitase and mitochondrial ironâ€“sulphur protein deficiency in Friedreich ataxia. Nature Genetics, 1997, 17, 215-217.	20.4	1,035
2	Mutation of a nuclear succinate dehydrogenase gene results in mitochondrial respiratory chain deficiency. Nature Genetics, 1995, 11, 144-149.	20.4	687
3	Persistent mitochondrial dysfunction and perinatal exposure to antiretroviral nucleoside analogues. Lancet, The, 1999, 354, 1084-1089.	12.1	628
4	Mutation of RRM2B, encoding p53-controlled ribonucleotide reductase (p53R2), causes severe mitochondrial DNA depletion. Nature Genetics, 2007, 39, 776-780.	20.4	483
5	Mitochondrial double-stranded RNA triggers antiviral signalling in humans. Nature, 2018, 560, 238-242.	36.2	445
6	Selective iron chelation in Friedreich ataxia: biologic and clinical implications. Blood, 2007, 110, 401-408.	1.4	412
7	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
8	Effect of idebenone on cardiomyopathy in Friedreich's ataxia: a preliminary study. Lancet, The, 1999, 354, 477-479.	12.1	355
9	COQ6 mutations in human patients produce nephrotic syndrome with sensorineural deafness. Journal of Clinical Investigation, 2011, 121, 2013-2024.	8.2	347
10	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
11	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
12	CABC1 Gene Mutations Cause Ubiquinone Deficiency with Cerebellar Ataxia and Seizures. American Journal of Human Genetics, 2008, 82, 623-630.	6.1	287
13	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
14	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
15	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
16	Succinate dehydrogenase and human diseases: new insights into a well-known enzyme. European Journal of Human Genetics, 2002, 10, 289-291.	2.9	216
17	Functional Consequences of aSDHBGene Mutation in an Apparently Sporadic Pheochromocytoma. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 4771-4774.	3.6	211
18	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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19	Spectrum of mitochondrial DNA rearrangements in the Pearson marrow-pancreas syndrome. Human Molecular Genetics, 1995, 4, 1327-1330.	3.0	196
20	Cytochrome oxidase in health and disease. Gene, 2002, 286, 53-63.	2.3	179
21	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
22	Twinkle helicase (PEO1) gene mutation causes mitochondrial DNA depletion. Annals of Neurology, 2007, 62, 579-587.	5.8	158
23	Epileptic phenotypes in children with respiratory chain disorders. Epilepsia, 2010, 51, 1225-1235.	4.6	155
24	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
25	Mutant NDUFV2 subunit of mitochondrial complex I causes early onset hypertrophic cardiomyopathy and encephalopathy. Human Mutation, 2003, 21, 582-586.	2.8	152
26	Assessment of the mitochondrial respiratory chain. Lancet, The, 1991, 338, 60.	12.1	151
27	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
28	Hepatic mitochondrial DNA deletion in alcoholics: Association with microvesicular steatosis. Gastroenterology, 1995, 108, 193-200.	1.4	146
29	Clinical presentations and laboratory investigations in respiratory chain deficiency. European Journal of Pediatrics, 1996, 155, 262-274.	2.7	137
30	Antenatal manifestations of mitochondrial respiratory chain deficiency. Journal of Pediatrics, 2003, 143, 208-212.	2.2	129
31	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
32	Human diseases with impaired mitochondrial protein synthesis. Biochimica Et Biophysica Acta - Bioenergetics, 2011, 1807, 1198-1205.	1.6	129
33	Exome sequencing identifies MRPL3 mutation in mitochondrial cardiomyopathy. Human Mutation, 2011, 32, 1225-1231.	2.8	128
34	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
35	Yeast as a system for modeling mitochondrial disease mechanisms and discovering therapies. DMM Disease Models and Mechanisms, 2015, 8, 509-526.	2.4	120
36	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

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37	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
38	Refractory epilepsy and mitochondrial dysfunction due to GM3 synthase deficiency. European Journal of Human Genetics, 2013, 21, 528-534.	2.9	110
39	Inborn errors of complex II â€“ Unusual human mitochondrial diseases. Biochimica Et Biophysica Acta - Bioenergetics, 2002, 1553, 117-122.	1.6	109
40	Site-specific deletions of the mitochondrial genome in the Pearson marrow-pancreas syndrome. Genomics, 1991, 10, 502-504.	2.9	108
41	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38.	8.5	107
42	Prevalence of rare mitochondrial DNA mutations in mitochondrial disorders. Journal of Medical Genetics, 2013, 50, 704-714.	3.6	105
43	Mitochondrial DNA rearrangements with onset as chronic diarrhea with villous atrophy. Journal of Pediatrics, 1994, 124, 63-70.	2.2	104
44	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
45	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
46	Coamplification of Nuclear Pseudogenes and Assessment of Heteroplasmy of Mitochondrial DNA Mutations. Biochemical and Biophysical Research Communications, 1998, 247, 57-59.	2.2	96
47	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
48	Neonatal and delayed-onset liver involvement in disorders of oxidative phosphorylation. Journal of Pediatrics, 1997, 130, 817-822.	2.2	94
49	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
50	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
51	Endomyocardial biopsies for early detection of mitochondrial disorders in hypertrophic cardiomyopathies. Journal of Pediatrics, 1994, 124, 224-228.	2.2	87
52	The kidney in mitochondrial cytopathies. Kidney International, 1997, 51, 1000-1007.	5.4	87
53	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
54	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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55	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
56	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
57	Heart Hypertrophy and Function Are Improved by Idebenone in Friedreich's Ataxia. Free Radical Research, 2002, 36, 467-469.	3.3	82
58	Deletion of the mitochondrial DNA in a case of de Toni-Debrï¿½-Fanconi syndrome and Pearson syndrome. Pediatric Nephrology, 1994, 8, 164-168.	1.8	80
59	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
60	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
61	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
62	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
63	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
64	Genetic causes of mitochondrial DNA depletion in humans. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2009, 1792, 1103-1108.	3.8	76
65	Impaired Transferrin Receptor Palmitoylation and Recycling in Neurodegeneration with Brain Iron Accumulation. American Journal of Human Genetics, 2018, 102, 266-277.	6.1	76
66	Genetic Features of Mitochondrial Respiratory Chain Disorders. Journal of the American Society of Nephrology: JASN, 2003, 14, 2995-3007.	0.5	75
67	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
68	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
69	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
70	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
71	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.	2.2	64
72	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

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73	Renal involvement in mitochondrial cytopathies. Pediatric Nephrology, 1996, 10, 368-373.	1.8	61
74	Hepatic failure in disorders of oxidative phosphorylation with neonatal onset. Journal of Pediatrics, 1991, 119, 951-954.	2.2	60
75	Infantile and pediatric quinone deficiency diseases. Mitochondrion, 2007, 7, S112-S121.	3.6	60
76	Molecular insights into Friedreich's ataxia and antioxidant-based therapies. Trends in Molecular Medicine, 2002, 8, 221-224.	7.1	56
77	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
78	NDUFS4 mutations cause Leigh syndrome with predominant brainstem involvement. Molecular Genetics and Metabolism, 2009, 97, 185-189.	2.2	55
79	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
80	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
81	DELETION OF BLOOD MITOCHONDRIAL DNA IN PANCYTOPENIA. Lancet, The, 1988, 332, 567-568.	12.1	53
82	Deletion of mitochondrial DNA in patient with chronic tubulointerstitial nephritis. Journal of Pediatrics, 1995, 126, 597-601.	2.2	52
83	Pearson Syndrome in the Neonatal Period. Journal of Pediatric Hematology/Oncology, 2009, 31, 947-951.	0.6	52
84	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
85	An improved spectrophotometric assay of pyruvate dehydrogenase in lactate dehydrogenase contaminated mitochondrial preparations from human skeletal muscle. Clinica Chimica Acta, 1995, 240, 129-136.	1.6	51
86	Fluxes of Nicotinamide Adenine Dinucleotides through Mitochondrial Membranes in Human Cultured Cells. Journal of Biological Chemistry, 1996, 271, 14785-14790.	3.5	50
87	Mitochondrial respiratory chain complex assembly and function during human fetal development. Molecular Genetics and Metabolism, 2008, 94, 120-126.	2.2	50
88	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
89	Respiratory chain deficiency presenting as congenital nephrotic syndrome. Pediatric Nephrology, 2005, 20, 465-469.	1.8	44
90	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

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91	Biallelic PPA2 Mutations Cause Sudden Unexpected Cardiac Arrest in Infancy. American Journal of Human Genetics, 2016, 99, 666-673.	6.1	42
92	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
93	Cerebral white matter disease in children may be caused by mitochondrial respiratory chain deficiency. Journal of Pediatrics, 2000, 136, 209-214.	2.2	41
94	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
95	Duplications of mitochondrial DNA in Kearns-Sayre syndrome. Muscle and Nerve, 1995, 18, S154-S158.	2.3	40
96	Idebenone treatment in Friedreich patients: One-year-long randomized placebo-controlled trial. Neurology, 2004, 62, 524-525.	1.1	40
97	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
98	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
99	A case of Pearson syndrome associated with multiple renal cysts. Pediatric Nephrology, 1996, 10, 637-638.	1.8	39
100	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
101	Respiratory Chain Deficiency in Alpers Syndrome. Neuropediatrics, 2001, 32, 150-152.	0.8	38
102	New SUCLG1 patients expanding the phenotypic spectrum of this rare cause of mild methylmalonic aciduria. Mitochondrion, 2010, 10, 335-341.	3.6	38
103	Molecular diagnostics of mitochondrial disorders. Biochimica Et Biophysica Acta - Bioenergetics, 2004, 1659, 129-135.	1.6	36
104	Mutation in the mitochondrial translation elongation factor EFTs results in severe infantile liver failure. Journal of Hepatology, 2012, 56, 294-297.	3.9	36
105	Genetic bases of mitochondrial respiratory chain disorders. Diabetes and Metabolism, 2010, 36, 97-107.	3.1	35
106	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
107	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
108	Loss of MTX2 causes mandibuloacral dysplasia and links mitochondrial dysfunction to altered nuclear morphology. Nature Communications, 2020, 11, 4589.	13.2	33

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109	Tubulopathy and pancytopaenia with normal pancreatic function: A variant of Pearson syndrome. European Journal of Medical Genetics, 2009, 52, 23-26.	1.3	32
110	Cytochrome c oxidase deficiency presenting as recurrent neonatal myoglobinuria. Neuromuscular Disorders, 1995, 5, 285-289.	0.7	31
111	Quinone analogs prevent enzymes targeted in Friedreich ataxia from ironâ€induced injury in vitro. BioFactors, 1999, 9, 247-251.	5.5	31
112	Macular dystrophy, diabetes, and deafness associated with a large mitochondrial dma deletion. American Journal of Ophthalmology, 1998, 125, 100-103.	3.4	30
113	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
114	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
115	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
116	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
117	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
118	Massive and exclusive pontocerebellar damage in mitochondrial disease and NUBPL mutations. Neurology, 2012, 79, 391-391.	1.1	27
119	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
120	Mouse models for mitochondrial diseases. Human Molecular Genetics, 2016, 25, R115-R122.	3.0	26
121	Expression of respiratory chain deficiencies in human cultured cells. Neuromuscular Disorders, 1993, 3, 605-608.	0.7	25
122	Liver cytochrome c oxidase deficiency in a case of neonatal-onset hepatic failure. European Journal of Pediatrics, 1994, 153, 190-194.	2.7	25
123	Haploinsufficiency of the GPD2 gene in a patient with nonsyndromic mental retardation. Human Genetics, 2009, 124, 649-658.	3.8	25
124	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
125	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
126	Toward genotype phenotype correlations in GFM1 mutations. Mitochondrion, 2012, 12, 242-247.	3.6	23

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127	Data from Artificial Models of Mitochondrial DNA Disorders Are Not Always Applicable to Humans. Cell Reports, 2014, 7, 933-934.	6.3	23
128	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
129	Another observation with VATER association and a complex IV respiratory chain deficiency. European Journal of Medical Genetics, 2006, 49, 71-77.	1.3	22
130	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
131	Mitochondrial Respiratory Chain Dysfunction Caused by Coenzyme Q Deficiency. Methods in Enzymology, 2004, 382, 81-88.	1.7	21
132	High predictive value of brain MRI imaging in primary mitochondrial respiratory chain deficiency. Journal of Medical Genetics, 2018, 55, 378-383.	3.6	21
133	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
134	Inherited glycosylphosphatidylinositol defects cause the rare Emm-negative blood phenotype and developmental disorders. Blood, 2021, 137, 3660-3669.	1.4	20
135	Defective palmitoylation of transferrin receptor triggers iron overload in Friedreich ataxia fibroblasts. Blood, 2021, 137, 2090-2102.	1.4	20
136	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
137	Expanding and Underscoring the Hepatoâ€Encephalopathic Phenotype of QIL1/MIC13. Hepatology, 2019, 70, 1066-1070.	8.1	19
138	No mitochondrial cytochrome oxidase (COX) gene mutations in 18 cases of COX deficiency. Human Genetics, 1997, 101, 247.	3.8	18
139	Respiratory chain deficiencies. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1651-1666.	0.3	18
140	Iron overload and mitochondrial diseases. Lancet, The, 1998, 351, 1286-1287.	12.1	17
141	Human cultured skin fibroblasts survive profound inherited ubiquinone depletion. Free Radical Research, 2001, 35, 11-21.	3.3	17
142	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
143	The natural history of infantile mitochondrial DNA depletion syndrome due to RRM2B deficiency. Genetics in Medicine, 2020, 22, 199-209.	2.4	17
144	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

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145	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
146	Respiratory Chain Deficiency Presenting as Recurrent Myoglobinuria in Childhood. Neuropediatrics, 1999, 30, 42-44.	0.8	13
147	Unusual presentation of Kearns-Sayre syndrome in early childhood. Pediatric Neurology, 1999, 21, 830-831.	2.1	13
148	Clinical, neuroimaging and biochemical findings in patients and patient fibroblasts expressing ten novel GFM1 mutations. Human Mutation, 2020, 41, 397-402.	2.8	13
149	Mitochondrial activities in human cultured skin fibroblasts contaminated by Mycoplasma hyorhinis. BMC Biochemistry, 2003, 4, 15.	4.3	11
150	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
151	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
152	Large Aperture Space Telescopes in Formation: Modeling, Metrology, and Control. Journal of the Astronautical Sciences, 2005, 53, 391-412.	1.4	11
153	Phenotypic expression of mitochondrial genotypes in cultured skin fibroblasts and in Epstein-Barr virus-transformed lymphocytes in Pearson syndrome. Muscle and Nerve, 1995, 18, S159-S164.	2.3	10
154	Heterozygous <scp>PNPT1</scp> Variants Cause Spinocerebellar Ataxia Type 25. Annals of Neurology, 2022, 92, 122-137.	5.8	10
155	Neonatal gene therapy achieves sustained disease rescue of maple syrup urine disease in mice. Nature Communications, 2022, 13, .	13.2	10
156	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
157	Secondary Mitochondrial Respiratory Chain Defect Can Delay Accurate PFIC2 Diagnosis. JIMD Reports, 2013, 14, 17-21.	1.2	8
158	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
159	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
160	Haematological characteristics and spontaneous haematological recovery in Pearson syndrome. British Journal of Haematology, 2021, 193, 1283-1287.	2.7	8
161	Defects of the Respiratory Chain. , 2012, , 223-238.		7
162	Pitfalls in molecular diagnosis of Friedreich ataxia. European Journal of Medical Genetics, 2018, 61, 455-458.	1.3	7

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163	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
164	Mitochondrial Complex I Deficiency in Humans. Current Genomics, 2004, 5, 137-146.	1.6	7
165	Screening Human EST Database for Identification of Candidate Genes in Respiratory Chain Deficiency. Molecular Genetics and Metabolism, 2000, 69, 223-232.	2.2	6
166	InstabilitÃ© du gÃ©nome mitochondrial et pathologies associÃ©es. Medecine/Sciences, 2010, 26, 171-176.	0.2	6
167	Heterogeneity of PNPT1 neuroimaging: mitochondriopathy, interferonopathy or both?. Journal of Medical Genetics, 2022, 59, 204-208.	3.6	6
168	Cerebral blood flow and acute episodes of Leigh syndrome in neurometabolic disorders. Developmental Medicine and Child Neurology, 2021, 63, 705-711.	2.7	6
169	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
170	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
171	News in Ubiquinone Biosynthesis. Chemistry and Biology, 2010, 17, 415-416.	6.2	3
172	Biallelic mutations in the <i>SARS2</i> gene presenting as congenital sideroblastic anemia. Haematologica, 2021, 106, 3202-3205.	3.5	3
173	Biallelic <i>IARS2</i> mutations presenting as sideroblastic anemia. Haematologica, 2021, 106, 0-0.	3.5	3
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