

Robert W Taylor

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/10610870/publications.pdf>

Version: 2024-02-01

351
papers

27,261
citations

7251

80
h-index

9865

146
g-index

356
all docs

356
docs citations

356
times ranked

24369
citing authors

#	ARTICLE	IF	CITATIONS
1	Mitochondrial DNA mutations in human disease. <i>Nature Reviews Genetics</i> , 2005, 6, 389-402.	7.7	1,530
2	High levels of mitochondrial DNA deletions in substantia nigra neurons in aging and Parkinson disease. <i>Nature Genetics</i> , 2006, 38, 515-517.	9.4	1,363
3	Prevalence of nuclear and mitochondrial <scp>DNA</scp> mutations related to adult mitochondrial disease. <i>Annals of Neurology</i> , 2015, 77, 753-759.	2.8	706
4	Prevalence of mitochondrial DNA disease in adults. <i>Annals of Neurology</i> , 2008, 63, 35-39.	2.8	540
5	Mitochondrial DNA mutations and human disease. <i>Biochimica Et Biophysica Acta - Bioenergetics</i> , 2010, 1797, 113-128.	0.5	527
6	Mitochondrial DNA mutations in human colonic crypt stem cells. <i>Journal of Clinical Investigation</i> , 2003, 112, 1351-1360.	3.9	454
7	Genetic diagnosis of Mendelian disorders via RNA sequencing. <i>Nature Communications</i> , 2017, 8, 15824.	5.8	432
8	Pronuclear transfer in human embryos to prevent transmission of mitochondrial DNA disease. <i>Nature</i> , 2010, 465, 82-85.	13.7	421
9	Phenotypic spectrum associated with mutations of the mitochondrial polymerase γ gene. <i>Brain</i> , 2006, 129, 1674-1684.	3.7	397
10	Mutation of OPA1 causes dominant optic atrophy with external ophthalmoplegia, ataxia, deafness and multiple mitochondrial DNA deletions: a novel disorder of mtDNA maintenance. <i>Brain</i> , 2008, 131, 329-337.	3.7	381
11	Universal heteroplasmy of human mitochondrial DNA. <i>Human Molecular Genetics</i> , 2013, 22, 384-390.	1.4	344
12	What causes mitochondrial DNA deletions in human cells?. <i>Nature Genetics</i> , 2008, 40, 275-279.	9.4	334
13	The genetics and pathology of mitochondrial disease. <i>Journal of Pathology</i> , 2017, 241, 236-250.	2.1	329
14	The epidemiology of mitochondrial disordersâ€”past, present and future. <i>Biochimica Et Biophysica Acta - Bioenergetics</i> , 2004, 1659, 115-120.	0.5	325
15	Biochemical Assays of Respiratory Chain Complex Activity. <i>Methods in Cell Biology</i> , 2007, 80, 93-119.	0.5	324
16	A neurological perspective on mitochondrial disease. <i>Lancet Neurology</i> , The, 2010, 9, 829-840.	4.9	316
17	Use of Whole-Exome Sequencing to Determine the Genetic Basis of Multiple Mitochondrial Respiratory Chain Complex Deficiencies. <i>JAMA - Journal of the American Medical Association</i> , 2014, 312, 68.	3.8	304
18	Detection and quantification of mitochondrial DNA deletions in individual cells by real-time PCR. <i>Nucleic Acids Research</i> , 2002, 30, 68e-68.	6.5	279

#	ARTICLE	IF	CITATIONS
19	Mitochondrial DNA mutations are established in human colonic stem cells, and mutated clones expand by crypt fission. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 714-719.	3.3	269
20	GRACILE Syndrome, a Lethal Metabolic Disorder with Iron Overload, Is Caused by a Point Mutation in BCS1L. American Journal of Human Genetics, 2002, 71, 863-876.	2.6	263
21	Selective inhibition of mutant human mitochondrial DNA replication in vitro by peptide nucleic acids. Nature Genetics, 1997, 15, 212-215.	9.4	252
22	Mutations causing mitochondrial disease: What is new and what challenges remain?. Science, 2015, 349, 1494-1499.	6.0	251
23	Mitochondrial dysfunction in a cell culture model of familial amyotrophic lateral sclerosis. Brain, 2002, 125, 1522-1533.	3.7	249
24	Mitochondrial Protein Interaction Mapping Identifies Regulators of Respiratory Chain Function. Molecular Cell, 2016, 63, 621-632.	4.5	241
25	Mitochondrial DNA and disease. Journal of Pathology, 2012, 226, 274-286.	2.1	239
26	Mechanisms of Field Cancerization in the Human Stomach: The Expansion and Spread of Mutated Gastric Stem Cells. Gastroenterology, 2008, 134, 500-510.	0.6	222
27	Recent Advances in Mitochondrial Disease. Annual Review of Genomics and Human Genetics, 2017, 18, 257-275.	2.5	217
28	Mutant POLG2 Disrupts DNA Polymerase γ Subunits and Causes Progressive External Ophthalmoplegia. American Journal of Human Genetics, 2006, 78, 1026-1034.	2.6	215
29	Superoxide dismutase downregulation in osteoarthritis progression and end-stage disease. Annals of the Rheumatic Diseases, 2010, 69, 1502-1510.	0.5	202
30	Risk of developing a mitochondrial DNA deletion disorder. Lancet, The, 2004, 364, 592-596.	6.3	201
31	mt $\langle scp \rangle$ DNA $\langle /scp \rangle$ heteroplasmy level and copy number indicate disease burden in m.3243A $\langle \>G$ mitochondrial disease. EMBO Molecular Medicine, 2018, 10, .	3.3	199
32	The UK MRC Mitochondrial Disease Patient Cohort Study: clinical phenotypes associated with the m.3243A $\langle \>G$ mutation—implications for diagnosis and management. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 936-938.	0.9	193
33	Endurance training and detraining in mitochondrial myopathies due to single large-scale mtDNA deletions. Brain, 2006, 129, 3391-3401.	3.7	189
34	Cardiac involvement in mitochondrial DNA disease: clinical spectrum, diagnosis, and management. European Heart Journal, 2012, 33, 3023-3033.	1.0	182
35	The diagnosis of mitochondrial muscle disease. Neuromuscular Disorders, 2004, 14, 237-245.	0.3	178
36	The Spectrum of Mitochondrial Ultrastructural Defects in Mitochondrial Myopathy. Scientific Reports, 2016, 6, 30610.	1.6	165

#	ARTICLE	IF	CITATIONS
37	De novo mutations in the mitochondrialND3 gene as a cause of infantile mitochondrial encephalopathy and complex I deficiency. <i>Annals of Neurology</i> , 2004, 55, 58-64.	2.8	164
38	Induction of Mitochondrial Biogenesis Is a Maladaptive Mechanism in Mitochondrial Cardiomyopathies. <i>Journal of the American College of Cardiology</i> , 2007, 50, 1362-1369.	1.2	164
39	NDUFS6 mutations are a novel cause of lethal neonatal mitochondrial complex I deficiency. <i>Journal of Clinical Investigation</i> , 2004, 114, 837-845.	3.9	164
40	Multiple neonatal deaths due to a homoplasmic mitochondrial DNA mutation. <i>Nature Genetics</i> , 2002, 30, 145-146.	9.4	162
41	A homoplasmic mitochondrial transfer Ribonucleic Acid mutation as a cause of maternally inherited hypertrophic cardiomyopathy. <i>Journal of the American College of Cardiology</i> , 2003, 41, 1786-1796.	1.2	161
42	Assigning pathogenicity to mitochondrial tRNA mutations: when "definitely maybe" is not good enough. <i>Trends in Genetics</i> , 2004, 20, 591-596.	2.9	159
43	A comparative analysis approach to determining the pathogenicity of mitochondrial tRNA mutations. <i>Human Mutation</i> , 2011, 32, 1319-1325.	1.1	159
44	Dominant inheritance of premature ovarian failure associated with mutant mitochondrial DNA polymerase gamma. <i>Human Reproduction</i> , 2006, 21, 2467-2473.	0.4	153
45	Mutations in the SPG7 gene cause chronic progressive external ophthalmoplegia through disordered mitochondrial DNA maintenance. <i>Brain</i> , 2014, 137, 1323-1336.	3.7	151
46	Resistance training in patients with single, large-scale deletions of mitochondrial DNA. <i>Brain</i> , 2008, 131, 2832-2840.	3.7	147
47	Mitochondrial tRNA mutations and disease. <i>Wiley Interdisciplinary Reviews RNA</i> , 2010, 1, 304-324.	3.2	145
48	Mutations in FBXL4 Cause Mitochondrial Encephalopathy and a Disorder of Mitochondrial DNA Maintenance. <i>American Journal of Human Genetics</i> , 2013, 93, 471-481.	2.6	137
49	Locating the stem cell niche and tracing hepatocyte lineages in human liver. <i>Hepatology</i> , 2009, 49, 1655-1663.	3.6	135
50	ELAC2 Mutations Cause a Mitochondrial RNA Processing Defect Associated with Hypertrophic Cardiomyopathy. <i>American Journal of Human Genetics</i> , 2013, 93, 211-223.	2.6	127
51	Late-onset optic atrophy, ataxia, and myopathy associated with a mutation of a complex II gene. <i>Annals of Neurology</i> , 2000, 48, 330-335.	2.8	126
52	Mutations in GTPBP3 Cause a Mitochondrial Translation Defect Associated with Hypertrophic Cardiomyopathy, Lactic Acidosis, and Encephalopathy. <i>American Journal of Human Genetics</i> , 2014, 95, 708-720.	2.6	123
53	The GABA Transaminase, ABAT, Is Essential for Mitochondrial Nucleoside Metabolism. <i>Cell Metabolism</i> , 2015, 21, 417-427.	7.2	119
54	Quantitative 3D Mapping of the Human Skeletal Muscle Mitochondrial Network. <i>Cell Reports</i> , 2019, 26, 996-1009.e4.	2.9	116

#	ARTICLE	IF	CITATIONS
55	An mtDNA Mutation in the Initiation Codon of the Cytochrome C Oxidase Subunit II Gene Results in Lower Levels of the Protein and a Mitochondrial Encephalomyopathy. <i>American Journal of Human Genetics</i> , 1999, 64, 1330-1339.	2.6	115
56	The neurology of mitochondrial DNA disease. <i>Lancet Neurology</i> , The, 2002, 1, 343-351.	4.9	115
57	Clonal Expansion of Early to Mid-Life Mitochondrial DNA Point Mutations Drives Mitochondrial Dysfunction during Human Ageing. <i>PLoS Genetics</i> , 2014, 10, e1004620.	1.5	115
58	Mitochondrial dysfunction in osteoarthritis is associated with downregulation of superoxide dismutase 2. <i>Arthritis and Rheumatism</i> , 2013, 65, 378-387.	6.7	113
59	SANDO syndrome in a cohort of 107 patients with CPEO and mitochondrial DNA deletions. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2015, 86, 630-634.	0.9	113
60	Recent advances in understanding the molecular genetic basis of mitochondrial disease. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 36-50.	1.7	113
61	Molecular basis of infantile reversible cytochrome c oxidase deficiency myopathy. <i>Brain</i> , 2009, 132, 3165-3174.	3.7	112
62	Defective i6A37 Modification of Mitochondrial and Cytosolic tRNAs Results from Pathogenic Mutations in TRIT1 and Its Substrate tRNA. <i>PLoS Genetics</i> , 2014, 10, e1004424.	1.5	112
63	Biallelic Mutations in NBAS Cause Recurrent Acute Liver Failure with Onset in Infancy. <i>American Journal of Human Genetics</i> , 2015, 97, 163-169.	2.6	110
64	Noninvasive diagnosis of the 3243A>G mitochondrial DNA mutation using urinary epithelial cells. <i>European Journal of Human Genetics</i> , 2004, 12, 778-781.	1.4	107
65	Cell-permeable succinate prodrugs bypass mitochondrial complex I deficiency. <i>Nature Communications</i> , 2016, 7, 12317.	5.8	106
66	A novel immunofluorescent assay to investigate oxidative phosphorylation deficiency in mitochondrial myopathy: understanding mechanisms and improving diagnosis. <i>Scientific Reports</i> , 2015, 5, 15037.	1.6	104
67	Disease progression in patients with single, large-scale mitochondrial DNA deletions. <i>Brain</i> , 2014, 137, 323-334.	3.7	103
68	Respiratory chain complex I deficiency caused by mitochondrial DNA mutations. <i>European Journal of Human Genetics</i> , 2011, 19, 769-775.	1.4	102
69	Topoisomerase 3 \pm Is Required for Decatenation and Segregation of Human mtDNA. <i>Molecular Cell</i> , 2018, 69, 9-23.e6.	4.5	102
70	Phenotypic heterogeneity in m.3243A>G mitochondrial disease: The role of nuclear factors. <i>Annals of Clinical and Translational Neurology</i> , 2018, 5, 333-345.	1.7	102
71	Recessive germline <i>SDHA</i> and <i>SDHB</i> mutations causing leukodystrophy and isolated mitochondrial complex II deficiency. <i>Journal of Medical Genetics</i> , 2012, 49, 569-577.	1.5	100
72	Mitochondrial disease in adults: recent advances and future promise. <i>Lancet Neurology</i> , The, 2021, 20, 573-584.	4.9	96

#	ARTICLE	IF	CITATIONS
73	Endocrine disorders in mitochondrial disease. <i>Molecular and Cellular Endocrinology</i> , 2013, 379, 2-11.	1.6	93
74	Recurrent De Novo Dominant Mutations in SLC25A4 Cause Severe Early-Onset Mitochondrial Disease and Loss of Mitochondrial DNA Copy Number. <i>American Journal of Human Genetics</i> , 2016, 99, 860-876.	2.6	93

75

#	ARTICLE	IF	CITATIONS
91	Succinate-CoA ligase deficiency due to mutations in <i>SUCLA2</i> and <i>SUCLG1</i> : phenotype and genotype correlations in 71 patients. <i>Journal of Inherited Metabolic Disease</i> , 2016, 39, 243-252.	1.7	79
92	A multiplex real-time PCR method to detect and quantify mitochondrial DNA deletions in individual cells. <i>Analytical Biochemistry</i> , 2007, 370, 127-129.	1.1	77
93	Impaired mitochondrial biogenesis is a common feature to myocardial hypertrophy and end-stage ischemic heart failure. <i>Cardiovascular Pathology</i> , 2016, 25, 103-112.	0.7	77
94	Genotypes from patients indicate no paternal mitochondrial DNA contribution. <i>Annals of Neurology</i> , 2003, 54, 521-524.	2.8	76
95	Overexpression of human mitochondrial valyl tRNA synthetase can partially restore levels of cognate mt-tRNA ^{Val} carrying the pathogenic C25U mutation. <i>Nucleic Acids Research</i> , 2008, 36, 3065-3074.	6.5	74
96	Mitochondrial OXPHOS Biogenesis: Co-Regulation of Protein Synthesis, Import, and Assembly Pathways. <i>International Journal of Molecular Sciences</i> , 2020, 21, 3820.	1.8	74
97	Mitochondrial and inflammatory changes in sporadic inclusion body myositis. <i>Neuropathology and Applied Neurobiology</i> , 2015, 41, 288-303.	1.8	73
98	Retrospective natural history of thymidine kinase 2 deficiency. <i>Journal of Medical Genetics</i> , 2018, 55, 515-521.	1.5	73
99	Depletion of mitochondrial DNA in leucocytes harbouring the 3243A->G mtDNA mutation. <i>Journal of Medical Genetics</i> , 2006, 44, 69-74.	1.5	72
100	A Methodological Approach to Tracing Cell Lineage in Human Epithelial Tissues. <i>Stem Cells</i> , 2009, 27, 1410-1420.	1.4	72
101	Familial myopathy: New insights into the T14709C mitochondrial tRNA mutation. <i>Annals of Neurology</i> , 2004, 55, 478-484.	2.8	71
102	Adults with RRM2B-related mitochondrial disease have distinct clinical and molecular characteristics. <i>Brain</i> , 2012, 135, 3392-3403.	3.7	70
103	Deficiency of complex II of the mitochondrial respiratory chain in late-onset optic atrophy and ataxia. <i>Annals of Neurology</i> , 1996, 39, 224-232.	2.8	69
104	The p.M292T <i>NDUFS2</i> mutation causes complex I-deficient Leigh syndrome in multiple families. <i>Brain</i> , 2010, 133, 2952-2963.	3.7	69
105	Clinical, Genetic, and Radiological Features of Extrapyrmidal Movement Disorders in Mitochondrial Disease. <i>JAMA Neurology</i> , 2016, 73, 668.	4.5	69
106	Age-associated mitochondrial DNA mutations cause metabolic remodeling that contributes to accelerated intestinal tumorigenesis. <i>Nature Cancer</i> , 2020, 1, 976-989.	5.7	69
107	Isoleucyl-tRNA synthetase levels modulate the penetrance of a homoplasmic m.4277T>C mitochondrial tRNA ^{Leu} mutation causing hypertrophic cardiomyopathy. <i>Human Molecular Genetics</i> , 2012, 21, 85-100.	1.4	67
108	<i>MTO1</i> Mutations are Associated with Hypertrophic Cardiomyopathy and Lactic Acidosis and Cause Respiratory Chain Deficiency in Humans and Yeast. <i>Human Mutation</i> , 2013, 34, 1501-1509.	1.1	67

#	ARTICLE	IF	CITATIONS
109	MPV17 Loss Causes Deoxynucleotide Insufficiency and Slow DNA Replication in Mitochondria. <i>PLoS Genetics</i> , 2016, 12, e1005779.	1.5	67
110	Nuclear factors involved in mitochondrial translation cause a subgroup of combined respiratory chain deficiency. <i>Brain</i> , 2011, 134, 183-195.	3.7	66
111	Accurate mitochondrial DNA sequencing using off-target reads provides a single test to identify pathogenic point mutations. <i>Genetics in Medicine</i> , 2014, 16, 962-971.	1.1	64
112	Mutations in APOPT1, Encoding a Mitochondrial Protein, Cause Cavitating Leukoencephalopathy with Cytochrome c Oxidase Deficiency. <i>American Journal of Human Genetics</i> , 2014, 95, 315-325.	2.6	64
113	Multipotent Basal Stem Cells, Maintained in Localized Proximal Niches, Support Directed Long-Ranging Epithelial Flows in Human Prostates. <i>Cell Reports</i> , 2017, 20, 1609-1622.	2.9	64
114	Mutations in MDH2, Encoding a Krebs Cycle Enzyme, Cause Early-Onset Severe Encephalopathy. <i>American Journal of Human Genetics</i> , 2017, 100, 151-159.	2.6	63
115	<i>In situ</i> lineage tracking of human prostatic epithelial stem cell fate reveals a common clonal origin for basal and luminal cells. <i>Journal of Pathology</i> , 2011, 225, 181-188.	2.1	62
116	Pathogenic Mitochondrial tRNA Point Mutations: Nine Novel Mutations Affirm Their Importance as a Cause of Mitochondrial Disease. <i>Human Mutation</i> , 2013, 34, 1260-1268.	1.1	62
117	Epilepsy in adults with mitochondrial disease: A cohort study. <i>Annals of Neurology</i> , 2015, 78, 949-957.	2.8	62
118	A novel ANT1 gene mutation with probable germline mosaicism in autosomal dominant progressive external ophthalmoplegia. <i>Neuromuscular Disorders</i> , 2005, 15, 311-315.	0.3	61
119	Comparison of Mitochondrial Mutation Spectra in Ageing Human Colonic Epithelium and Disease: Absence of Evidence for Purifying Selection in Somatic Mitochondrial DNA Point Mutations. <i>PLoS Genetics</i> , 2012, 8, e1003082.	1.5	61
120	Mutation of the human mitochondrial phenylalanine-tRNA synthetase causes infantile-onset epilepsy and cytochrome c oxidase deficiency. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2014, 1842, 56-64.	1.8	61
121	Sensory neuronopathy in patients harbouring recessive polymerase β mutations. <i>Brain</i> , 2012, 135, 62-71.	3.7	60
122	Treatment of mitochondrial disease. <i>Journal of Bioenergetics and Biomembranes</i> , 1997, 29, 195-205.	1.0	59
123	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. <i>American Journal of Human Genetics</i> , 2018, 102, 494-504.	2.6	59
124	Targeting large molecules to mitochondria. <i>Advanced Drug Delivery Reviews</i> , 2001, 49, 189-198.	6.6	58
125	A novel sporadic mutation in cytochrome c oxidase subunit II as a cause of rhabdomyolysis. <i>Neuromuscular Disorders</i> , 2004, 14, 162-166.	0.3	58
126	Mitochondrial DNA Defects and Selective Extraocular Muscle Involvement in CPEO. , 2010, 51, 3340.		58

#	ARTICLE	IF	CITATIONS
127	Biallelic C1QBP Mutations Cause Severe Neonatal-, Childhood-, or Later-Onset Cardiomyopathy Associated with Combined Respiratory-Chain Deficiencies. <i>American Journal of Human Genetics</i> , 2017, 101, 525-538.	2.6	58
128	Neonatal Fanconi syndrome due to deficiency of complex III of the respiratory chain. <i>Pediatric Nephrology</i> , 1995, 9, 407-411.	0.9	57
129	A national perspective on prenatal testing for mitochondrial disease. <i>European Journal of Human Genetics</i> , 2014, 22, 1255-1259.	1.4	57
130	Biallelic Mutations in TMEM126B Cause Severe Complex I Deficiency with a Variable Clinical Phenotype. <i>American Journal of Human Genetics</i> , 2016, 99, 217-227.	2.6	57
131	Succinate-cytochrome c reductase: assessment of its value in the investigation of defects of the respiratory chain. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 1993, 1181, 261-265.	1.8	56
132	Age-associated mitochondrial DNA mutations lead to small but significant changes in cell proliferation and apoptosis in human colonic crypts. <i>Aging Cell</i> , 2010, 9, 96-99.	3.0	56
133	Nucleotide pools dictate the identity and frequency of ribonucleotide incorporation in mitochondrial DNA. <i>PLoS Genetics</i> , 2017, 13, e1006628.	1.5	55
134	Motor neuron disease in a patient with a mitochondrial tRNA ^{Ala} mutation. <i>Annals of Neurology</i> , 2006, 59, 570-574.	2.8	54
135	De novo mtDNA point mutations are common and have a low recurrence risk. <i>Journal of Medical Genetics</i> , 2017, 54, 73-83.	1.5	54
136	<i>OXAL</i> mutations cause mitochondrial encephalopathy and a combined oxidative phosphorylation defect. <i>EMBO Molecular Medicine</i> , 2018, 10, .	3.3	54
137	Mitochondrial DNA sequence characteristics modulate the size of the genetic bottleneck. <i>Human Molecular Genetics</i> , 2016, 25, 1031-1041.	1.4	53
138	Sudden adult death syndrome in m.3243A>G-related mitochondrial disease: an unrecognized clinical entity in young, asymptomatic adults. <i>European Heart Journal</i> , 2016, 37, 2552-2559.	1.0	53
139	Accurate Measurement of Mitochondrial DNA Deletion Level and Copy Number Differences in Human Skeletal Muscle. <i>PLoS ONE</i> , 2014, 9, e114462.	1.1	53
140	Defects in multiple complexes of the respiratory chain are present in ageing human colonic crypts. <i>Experimental Gerontology</i> , 2010, 45, 573-579.	1.2	52
141	Clinical, biochemical, cellular and molecular characterization of mitochondrial DNA depletion syndrome due to novel mutations in the MPV17 gene. <i>European Journal of Human Genetics</i> , 2014, 22, 184-191.	1.4	52
142	MPV17 mutation causes neuropathy and leukoencephalopathy with multiple mtDNA deletions in muscle. <i>Neuromuscular Disorders</i> , 2012, 22, 587-591.	0.3	51
143	NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. <i>Brain</i> , 2019, 142, 50-58.	3.7	51
144	SCYL1 variants cause a syndrome with low γ -glutamyl-transferase cholestasis, acute liver failure, and neurodegeneration (CALFAN). <i>Genetics in Medicine</i> , 2018, 20, 1255-1265.	1.1	50

#	ARTICLE	IF	CITATIONS
145	Pathogenic mitochondrial tRNA mutations - Which mutations are inherited and why?. Human Mutation, 2009, 30, E984-E992.	1.1	49
146	A recessive homozygous p.Asp92Gly SDHD mutation causes prenatal cardiomyopathy and a severe mitochondrial complex II deficiency. Human Genetics, 2015, 134, 869-879.	1.8	49
147	Defining mitochondrial protein functions through deep multiomic profiling. Nature, 2022, 606, 382-388.	13.7	49
148	Changes in the human mitochondrial genome after treatment of malignant disease. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2003, 525, 19-27.	0.4	48
149	Somatic Mitochondrial DNA Deletions Accumulate to High Levels in Aging Human Extraocular Muscles. , 2010, 51, 3347.		48
150	Evidence of severe mitochondrial oxidative stress and a protective effect of low oxygen in mouse models of inherited photoreceptor degeneration. Human Molecular Genetics, 2011, 20, 322-335.	1.4	48
151	Ultrasensitive deletion detection links mitochondrial DNA replication, disease, and aging. Genome Biology, 2020, 21, 248.	3.8	48
152	Subcellular origin of mitochondrial DNA deletions in human skeletal muscle. Annals of Neurology, 2018, 84, 289-301.	2.8	47
153	What is influencing the phenotype of the common homozygous polymerase- β mutation p.Ala467Thr?. Brain, 2012, 135, 3614-3626.	3.7	46
154	Structural modeling of tissue-specific mitochondrial alanyl-tRNA synthetase (AARS2) defects predicts differential effects on aminoacylation. Frontiers in Genetics, 2015, 6, 21.	1.1	46
155	Clinical, morphological, biochemical, imaging and outcome parameters in 21 individuals with mitochondrial maintenance defect related to <i>FBXL4</i> mutations. Journal of Inherited Metabolic Disease, 2015, 38, 905-914.	1.7	45
156	Metabolic effects of bezafibrate in mitochondrial disease. EMBO Molecular Medicine, 2020, 12, e11589.	3.3	45
157	Microangiopathy in the cerebellum of patients with mitochondrial DNA disease. Brain, 2012, 135, 1736-1750.	3.7	44
158	Clinical and Molecular Characteristics of Mitochondrial DNA Depletion Syndrome Associated with Neonatal Cholestasis and Liver Failure. Journal of Pediatrics, 2014, 164, 553-559.e2.	0.9	44
159	<i>FBXL4</i> deficiency increases mitochondrial removal by autophagy. EMBO Molecular Medicine, 2020, 12, e11659.	3.3	44
160	Defining cardiac adaptations and safety of endurance training in patients with m.3243A>G-related mitochondrial disease. International Journal of Cardiology, 2013, 168, 3599-3608.	0.8	43
161	The isolated carboxy-terminal domain of human mitochondrial leucyl-tRNA synthetase rescues the pathological phenotype of mitochondrial <i>tRNA</i> mutations in human cells. EMBO Molecular Medicine, 2014, 6, 169-182.	3.3	43
162	A novel inborn error of the coenzyme Q10 biosynthesis pathway: cerebellar ataxia and static encephalomyopathy due to COQ5 methyltransferase deficiency. Human Mutation, 2018, 39, 69-79.	1.1	43

#	ARTICLE	IF	CITATIONS
163	Clinical presentation and proteomic signature of patients with <i>TANGO2</i> mutations. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 297-308.	1.7	43
164	Developmental and Pathological Changes in the Human Cardiac Muscle Mitochondrial DNA Organization, Replication and Copy Number. <i>PLoS ONE</i> , 2010, 5, e10426.	1.1	43
165	Novel mutations in the TK2 gene associated with fatal mitochondrial DNA depletion myopathy. <i>Neuromuscular Disorders</i> , 2008, 18, 557-560.	0.3	42
166	POLG1 Mutations Manifesting as Autosomal Recessive Axonal Charcot-Marie-Tooth Disease. <i>Archives of Neurology</i> , 2008, 65, 133-6.	4.9	42
167	A New Mitochondrial Transfer RNA Pro Gene Mutation Associated With Myoclonic Epilepsy With Ragged-Red Fibers and Other Neurological Features. <i>Archives of Neurology</i> , 2009, 66, 399-402.	4.9	42
168	Adult-onset spinocerebellar ataxia syndromes due to <i>MTATP6</i> mutations. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2012, 83, 883-886.	0.9	42
169	Pathological mechanisms underlying single large-scale mitochondrial DNA deletions. <i>Annals of Neurology</i> , 2018, 83, 115-130.	2.8	42
170	A novel Twinkle gene mutation in autosomal dominant progressive external ophthalmoplegia. <i>Neuromuscular Disorders</i> , 2003, 13, 568-572.	0.3	41
171	Clonal Expansion of Secondary Mitochondrial DNA Deletions Associated With Spinocerebellar Ataxia Type 28. <i>JAMA Neurology</i> , 2015, 72, 106.	4.5	41
172	The urinary proteome and metabolome differ from normal in adults with mitochondrial disease. <i>Kidney International</i> , 2015, 87, 610-622.	2.6	41
173	Clinical Features, Molecular Heterogeneity, and Prognostic Implications in <i>YARS2</i> -Related Mitochondrial Myopathy. <i>JAMA Neurology</i> , 2017, 74, 686.	4.5	41
174	Bi-allelic Mutations in NDUFA6 Establish Its Role in Early-Onset Isolated Mitochondrial Complex I Deficiency. <i>American Journal of Human Genetics</i> , 2018, 103, 592-601.	2.6	41
175	Incidence of Primary Mitochondrial Disease in Children Younger Than 2 Years Presenting With Acute Liver Failure. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2016, 63, 592-597.	0.9	40
176	Pseudo-obstruction, stroke, and mitochondrial dysfunction: A lethal combination. <i>Annals of Neurology</i> , 2016, 80, 686-692.	2.8	40
177	Clonally Expanded Mitochondrial DNA Mutations in Epileptic Individuals With Mutated DNA Polymerase β . <i>Journal of Neuropathology and Experimental Neurology</i> , 2008, 67, 857-866.	0.9	39
178	Cytochrome c oxidase-intermediate fibres: Importance in understanding the pathogenesis and treatment of mitochondrial myopathy. <i>Neuromuscular Disorders</i> , 2012, 22, 690-698.	0.3	39
179	Pathogenic Bi-allelic Mutations in NDUFAF8 Cause Leigh Syndrome with an Isolated Complex I Deficiency. <i>American Journal of Human Genetics</i> , 2020, 106, 92-101.	2.6	39
180	Mitochondrial DNA mutations in human disease. <i>IUBMB Life</i> , 2006, 58, 143-151.	1.5	37

#	ARTICLE	IF	CITATIONS
181	Mitochondrial pathology in progressive cerebellar ataxia. <i>Cerebellum and Ataxias</i> , 2015, 2, 16.	1.9	37
182	Complex mitochondrial DNA rearrangements in individual cells from patients with sporadic inclusion body myositis. <i>Nucleic Acids Research</i> , 2016, 44, 5313-5329.	6.5	37
183	Clinical, biochemical, and genetic features of four patients with short-chain enoyl-CoA hydratase (ECHS1) deficiency. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1115-1127.	0.7	36
184	The clinical spectrum of the m.10191T>C mutation in complex I-deficient Leigh syndrome. <i>Developmental Medicine and Child Neurology</i> , 2012, 54, 500-506.	1.1	35
185	The role of the mitochondrial ribosome in human disease: searching for mutations in 12S mitochondrial rRNA with high disruptive potential. <i>Human Molecular Genetics</i> , 2014, 23, 949-967.	1.4	35
186	The clinical, biochemical and genetic features associated with <i>RMND1</i> -related mitochondrial disease. <i>Journal of Medical Genetics</i> , 2016, 53, 768-775.	1.5	35
187	A <i>Wars2</i> Mutant Mouse Model Displays OXPHOS Deficiencies and Activation of Tissue-Specific Stress Response Pathways. <i>Cell Reports</i> , 2018, 25, 3315-3328.e6.	2.9	35
188	A novel mitochondrial <i>MTND5</i> frameshift mutation causing isolated complex I deficiency, renal failure and myopathy. <i>Neuromuscular Disorders</i> , 2010, 20, 131-135.	0.3	34
189	<i>Bmi1</i> enhances skeletal muscle regeneration through <i>MT1</i> -mediated oxidative stress protection in a mouse model of dystrophinopathy. <i>Journal of Experimental Medicine</i> , 2014, 211, 2617-2633.	4.2	34
190	Dissecting the mechanisms underlying the accumulation of mitochondrial DNA deletions in human skeletal muscle. <i>Human Molecular Genetics</i> , 2014, 23, 4612-4620.	1.4	34
191	Copy-choice recombination during mitochondrial L-strand synthesis causes DNA deletions. <i>Nature Communications</i> , 2019, 10, 759.	5.8	34
192	<i>POLG</i> mutations cause decreased mitochondrial DNA repopulation rates following induced depletion in human fibroblasts. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2011, 1812, 321-325.	1.8	33
193	Mitochondrial DNA deletions in muscle satellite cells: implications for therapies. <i>Human Molecular Genetics</i> , 2013, 22, 4739-4747.	1.4	33
194	Pathogenic variants in <i>MT-ATP6</i> : A United Kingdom-based mitochondrial disease cohort study. <i>Annals of Neurology</i> , 2019, 86, 310-315.	2.8	33
195	The genetics of mitochondrial disease: dissecting mitochondrial pathology using multi-omic pipelines. <i>Journal of Pathology</i> , 2021, 254, 430-442.	2.1	33
196	Novel Mitochondrial Transfer RNAPhe Gene Mutation Associated With Late-Onset Neuromuscular Disease. <i>Archives of Neurology</i> , 2006, 63, 902.	4.9	32
197	A homoplasmic mtDNA variant can influence the phenotype of the pathogenic m.7472Cins <i>MTTS1</i> mutation: are two mutations better than one?. <i>European Journal of Human Genetics</i> , 2008, 16, 1265-1274.	1.4	32
198	The m.5650G>A mitochondrial tRNA ^{Ala} mutation is pathogenic and causes a phenotype of pure myopathy. <i>Neuromuscular Disorders</i> , 2008, 18, 63-67.	0.3	32

#	ARTICLE	IF	CITATIONS
199	Cardiomyopathies due to homoplasmic mitochondrial tRNA mutations: morphologic and molecular features. <i>Human Pathology</i> , 2013, 44, 1262-1270.	1.1	32
200	A truncating PET100 variant causing fatal infantile lactic acidosis and isolated cytochrome c oxidase deficiency. <i>European Journal of Human Genetics</i> , 2015, 23, 935-939.	1.4	32
201	Mitochondrial dysfunction in myofibrillar myopathy. <i>Neuromuscular Disorders</i> , 2016, 26, 691-701.	0.3	32
202	Assessment of mitochondrial respiratory chain enzymes in cells and tissues. <i>Methods in Cell Biology</i> , 2020, 155, 121-156.	0.5	32
203	Differences in RNA processing underlie the tissue specific phenotype of ISCU myopathy. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2010, 1802, 539-544.	1.8	31
204	Neuropathologic Characterization of Pontocerebellar Hypoplasia Type 6 Associated With Cardiomyopathy and Hydrops Fetalis and Severe Multisystem Respiratory Chain Deficiency due to Novel <i>RARS2</i> Mutations. <i>Journal of Neuropathology and Experimental Neurology</i> , 2015, 74, 688-703.	0.9	31
205	A recurrent mitochondrial p.Trp22ArgNDUFB3 variant causes a distinctive facial appearance, short stature and a mild biochemical and clinical phenotype. <i>Journal of Medical Genetics</i> , 2016, 53, 634-641.	1.5	31
206	Mitochondrial oxodicarboxylate carrier deficiency is associated with mitochondrial DNA depletion and spinal muscular atrophy-like disease. <i>Genetics in Medicine</i> , 2018, 20, 1224-1235.	1.1	31
207	Mutations in <i>ELAC2</i> associated with hypertrophic cardiomyopathy impair mitochondrial tRNA 3' end processing. <i>Human Mutation</i> , 2019, 40, 1731-1748.	1.1	31
208	An antigenomic strategy for treating heteroplasmic mtDNA disorders. <i>Advanced Drug Delivery Reviews</i> , 2001, 49, 121-125.	6.6	30
209	Characterization of mtDNA variation in a cohort of South African paediatric patients with mitochondrial disease. <i>European Journal of Human Genetics</i> , 2012, 20, 650-656.	1.4	30
210	Concentric hypertrophic remodelling and subendocardial dysfunction in mitochondrial DNA point mutation carriers. <i>European Heart Journal Cardiovascular Imaging</i> , 2013, 14, 650-658.	0.5	30
211	Triplex real-time PCR an improved method to detect a wide spectrum of mitochondrial DNA deletions in single cells. <i>Scientific Reports</i> , 2015, 5, 9906.	1.6	30
212	Preferential amplification of a human mitochondrial DNA deletion in vitro and in vivo. <i>Scientific Reports</i> , 2018, 8, 1799.	1.6	30
213	Mutations of the mitochondrial carrier translocase channel subunit TIM22 cause early-onset mitochondrial myopathy. <i>Human Molecular Genetics</i> , 2018, 27, 4135-4144.	1.4	30
214	The presence of highly disruptive 16S rRNA mutations in clinical samples indicates a wider role for mutations of the mitochondrial ribosome in human disease. <i>Mitochondrion</i> , 2015, 25, 17-27.	1.6	29
215	The molecular pathology of pathogenic mitochondrial tRNA variants. <i>FEBS Letters</i> , 2021, 595, 1003-1024.	1.3	29
216	Strategies for treating disorders of the mitochondrial genome. <i>Biochimica Et Biophysica Acta - Bioenergetics</i> , 2004, 1659, 232-239.	0.5	28

#	ARTICLE	IF	CITATIONS
217	A proposed consensus panel of organisms for determining evolutionary conservation of mt-tRNA point mutations. <i>Mitochondrion</i> , 2012, 12, 533-538.	1.6	28
218	Quantitative quadruple-label immunofluorescence of mitochondrial and cytoplasmic proteins in single neurons from human midbrain tissue. <i>Journal of Neuroscience Methods</i> , 2014, 232, 143-149.	1.3	28
219	Dysferlin mutations and mitochondrial dysfunction. <i>Neuromuscular Disorders</i> , 2016, 26, 782-788.	0.3	28
220	Experimental Strategies Towards Treating Mitochondrial DNA Disorders. <i>Bioscience Reports</i> , 2007, 27, 139-150.	1.1	27
221	Long-term survival of neonatal mitochondrial complex III deficiency associated with a novel BCS1L gene mutation. <i>Molecular Genetics and Metabolism</i> , 2010, 100, 345-348.	0.5	27
222	Extraocular Muscle Atrophy and Central Nervous System Involvement in Chronic Progressive External Ophthalmoplegia. <i>PLoS ONE</i> , 2013, 8, e75048.	1.1	27
223	Confirming TDP2 mutation in spinocerebellar ataxia autosomal recessive 23 (SCAR23). <i>Neurology: Genetics</i> , 2018, 4, e262.	0.9	27
224	The low abundance of clonally expanded mitochondrial DNA point mutations in aged substantia nigra neurons. <i>Aging Cell</i> , 2009, 8, 496-498.	3.0	26
225	Mitochondrial stress response triggered by defects in protein synthesis quality control. <i>Life Science Alliance</i> , 2019, 2, e201800219.	1.3	26
226	The investigation and diagnosis of pathogenic mitochondrial DNA mutations in human urothelial cells. <i>Biochemical and Biophysical Research Communications</i> , 2010, 393, 740-745.	1.0	25
227	Loss of Myelin-Associated Glycoprotein in Kearns-Sayre Syndrome. <i>Archives of Neurology</i> , 2012, 69, 490.	4.9	25
228	Clinical and biochemical features associated with <i>BCS1L</i> mutation. <i>Journal of Inherited Metabolic Disease</i> , 2013, 36, 813-820.	1.7	25
229	Scientific and Ethical Issues in Mitochondrial Donation. <i>New Bioethics</i> , 2018, 24, 57-73.	0.5	25
230	Resolving complexity in mitochondrial disease: Towards precision medicine. <i>Molecular Genetics and Metabolism</i> , 2019, 128, 19-29.	0.5	25
231	Forecasting stroke-like episodes and outcomes in mitochondrial disease. <i>Brain</i> , 2022, 145, 542-554.	3.7	25
232	Pathogenic mitochondrial mt-tRNA ^{Ala} variants are uniquely associated with isolated myopathy. <i>European Journal of Human Genetics</i> , 2015, 23, 1735-1738.	1.4	24
233	The genotypic and phenotypic spectrum of MTO1 deficiency. <i>Molecular Genetics and Metabolism</i> , 2018, 123, 28-42.	0.5	24
234	In-vitro genetic modification of mitochondrial function. <i>Human Reproduction</i> , 2000, 15, 79-85.	0.4	23

#	ARTICLE	IF	CITATIONS
235	Ophthalmoplegia due to mitochondrial DNA disease: The need for genetic diagnosis. <i>Muscle and Nerve</i> , 2005, 32, 104-107.	1.0	23
236	Late-onset respiratory failure due to <i>TK2</i> mutations causing multiple mtDNA deletions. <i>Neurology</i> , 2013, 81, 2051-2053.	1.5	23
237	Investigating complex <i>IV</i> deficiency in <i>P</i> urkinje cells and synapses in patients with mitochondrial disease. <i>Neuropathology and Applied Neurobiology</i> , 2016, 42, 477-492.	1.8	23
238	Lethal Neonatal LTBL Associated with Biallelic <i>EARS2</i> Variants: Case Report and Review of the Reported Neuroradiological Features. <i>JIMD Reports</i> , 2016, 33, 61-68.	0.7	23
239	Pathogenic variants in <i>HTRA2</i> cause an early-onset mitochondrial syndrome associated with 3-methylglutaconic aciduria. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 121-130.	1.7	23
240	Adult Onset Leigh Syndrome in the Intensive Care Setting: A Novel Presentation of a <i>C12orf65</i> Related Mitochondrial Disease. <i>Journal of Neuromuscular Diseases</i> , 2015, 2, 409-419.	1.1	22
241	Three families with <i>de novo</i> m.3243A>G mutation. <i>BBA Clinical</i> , 2016, 6, 19-24.	4.1	22
242	Clinical, biochemical, and genetic features associated with <i>VAR2</i> -related mitochondrial disease. <i>Human Mutation</i> , 2018, 39, 563-578.	1.1	22
243	The genetic basis of isolated mitochondrial complex II deficiency. <i>Molecular Genetics and Metabolism</i> , 2020, 131, 53-65.	0.5	22
244	Production of transmitochondrial cybrids containing naturally occurring pathogenic mtDNA variants. <i>Nucleic Acids Research</i> , 2006, 34, e95-e95.	6.5	21
245	A p.R369G <i>POLG2</i> mutation associated with adPEO and multiple mtDNA deletions causes decreased affinity between polymerase β subunits. <i>Mitochondrion</i> , 2012, 12, 313-319.	1.6	21
246	Novel <i>MTND1</i> mutations cause isolated exercise intolerance, complex I deficiency and increased assembly factor expression. <i>Clinical Science</i> , 2015, 128, 895-904.	1.8	21
247	Loss-of-function mutations in <i>ISCA2</i> disrupt 4Fe-4S cluster machinery and cause a fatal leukodystrophy with hyperglycinemia and mtDNA depletion. <i>Human Mutation</i> , 2018, 39, 537-549.	1.1	21
248	<i>POLRMT</i> mutations impair mitochondrial transcription causing neurological disease. <i>Nature Communications</i> , 2021, 12, 1135.	5.8	21
249	A novel mitochondrial DNA tRNA ^{Leu} (A4267G) mutation in a sporadic patient with mitochondrial myopathy. <i>Neuromuscular Disorders</i> , 2002, 12, 659-664.	0.3	20
250	A novel mitochondrial <i>ND5</i> (<i>MTND5</i>) gene mutation giving isolated exercise intolerance. <i>Neuromuscular Disorders</i> , 2008, 18, 310-314.	0.3	20
251	Maternally inherited mitochondrial DNA disease in consanguineous families. <i>European Journal of Human Genetics</i> , 2011, 19, 1226-1229.	1.4	20
252	Using a quantitative quadruple immunofluorescent assay to diagnose isolated mitochondrial Complex I deficiency. <i>Scientific Reports</i> , 2017, 7, 15676.	1.6	20

#	ARTICLE	IF	CITATIONS
253	Expanding the phenotype of de novo <i>SLC25A4</i> -linked mitochondrial disease to include mild myopathy. <i>Neurology: Genetics</i> , 2018, 4, e256.	0.9	20
254	Adult-onset Mendelian PEO Associated with Mitochondrial Disease. <i>Journal of Neuromuscular Diseases</i> , 2014, 1, 119-133.	1.1	19
255	Short peptides from leucyl-tRNA synthetase rescue disease-causing mitochondrial tRNA point mutations. <i>Human Molecular Genetics</i> , 2016, 25, 903-915.	1.4	19
256	Instability of the mitochondrial alanyl-tRNA synthetase underlies fatal infantile-onset cardiomyopathy. <i>Human Molecular Genetics</i> , 2019, 28, 258-268.	1.4	19
257	Molecular genetic investigations identify new clinical phenotypes associated with <i>BCS1L</i> -related mitochondrial disease. <i>Human Molecular Genetics</i> , 2019, 28, 3766-3776.	1.4	19
258	Height as a Clinical Biomarker of Disease Burden in Adult Mitochondrial Disease. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2019, 104, 2057-2066.	1.8	19
259	The mitochondrial genome and mitochondrial muscle disorders. <i>Current Opinion in Pharmacology</i> , 2001, 1, 288-293.	1.7	18
260	Childhood neurological presentation of a novel mitochondrial tRNA ^{Val} gene mutation. <i>Journal of the Neurological Sciences</i> , 2004, 225, 99-103.	0.3	18
261	Sporadic mitochondrial myopathy due to a new mutation in the mitochondrial tRNA ^{Ser} (UCN) gene. <i>Neuromuscular Disorders</i> , 2004, 14, 417-420.	0.3	18
262	Inosine Triphosphate Pyrophosphohydrolase (ITPA) polymorphic sequence variants in adult hematological malignancy patients and possible association with mitochondrial DNA defects. <i>Journal of Hematology and Oncology</i> , 2013, 6, 24.	6.9	18
263	Decreased male reproductive success in association with mitochondrial dysfunction. <i>European Journal of Human Genetics</i> , 2017, 25, 1162-1164.	1.4	18
264	Early-Onset Cataracts, Spastic Paraparesis, and Ataxia Caused by a Novel Mitochondrial tRNA ^{Glu} (MT-TE) Gene Mutation Causing Severe Complex I Deficiency: A Clinical, Molecular, and Neuropathologic Study. <i>Journal of Neuropathology and Experimental Neurology</i> , 2013, 72, 164-175.	0.9	17
265	Leigh syndrome caused by mutations in <i>MTFMT</i> is associated with a better prognosis. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 515-524.	1.7	17
266	Biallelic pathogenic variants in <i>NDUFC2</i> cause early-onset Leigh syndrome and stalled biogenesis of complex I. <i>EMBO Molecular Medicine</i> , 2020, 12, e12619.	3.3	17
267	Natural History of Leigh Syndrome: A Study of Disease Burden and Progression. <i>Annals of Neurology</i> , 2022, 91, 117-130.	2.8	17
268	Sensory ataxic neuropathy dysarthria and ophthalmoparesis (SANDO) in a sibling pair with a homozygous p.A467T <i>POLG</i> mutation. <i>Muscle and Nerve</i> , 2010, 41, 265-269.	1.0	16
269	Isolated Distal Myopathy of the Upper Limbs Associated With Mitochondrial DNA Depletion and Polymerase β Mutations. <i>Archives of Neurology</i> , 2010, 67, 1144-6.	4.9	16
270	Expanding the clinical phenotype of <i>IARS2</i> -related mitochondrial disease. <i>BMC Medical Genetics</i> , 2018, 19, 196.	2.1	16

#	ARTICLE	IF	CITATIONS
271	Understanding mitochondrial DNA maintenance disorders at the single muscle fibre level. <i>Nucleic Acids Research</i> , 2019, 47, 7430-7443.	6.5	16
272	Sporadic Intragenic Inversion of the Mitochondrial DNA MTND1 Gene Causing Fatal Infantile Lactic Acidosis. <i>Pediatric Research</i> , 2006, 59, 440-444.	1.1	15
273	Sporadic myopathy and exercise intolerance associated with the mitochondrial 8328G>A tRNALys mutation. <i>Journal of Neurology</i> , 2007, 254, 1283-1285.	1.8	15
274	Compound heterozygous RMND1 gene variants associated with chronic kidney disease, dilated cardiomyopathy and neurological involvement: a case report. <i>BMC Research Notes</i> , 2016, 9, 325.	0.6	15
275	Both mitochondrial DNA and mitonuclear gene mutations cause hearing loss through cochlear dysfunction. <i>Brain</i> , 2016, 139, e33-e33.	3.7	15
276	Clinically proven mtDNA mutations are not common in those with chronic fatigue syndrome. <i>BMC Medical Genetics</i> , 2017, 18, 29.	2.1	15
277	Mitochondrial DNA Transcription: Regulating the Power Supply. <i>Cell</i> , 2007, 130, 211-213.	13.5	14
278	Mitochondrial respiratory chain disease in children undergoing cardiac transplantation: A prospective study. <i>International Journal of Cardiology</i> , 2012, 155, 305-306.	0.8	14
279	The natural history of infantile mitochondrial DNA depletion syndrome due to RRM2B deficiency. <i>Genetics in Medicine</i> , 2020, 22, 199-209.	1.1	14
280	Novel MT-ND Gene Variants Causing Adult-Onset Mitochondrial Disease and Isolated Complex I Deficiency. <i>Frontiers in Genetics</i> , 2020, 11, 24.	1.1	14
281	Long-term survival in a child with severe encephalopathy, multiple respiratory chain deficiency and CFM1 mutations. <i>Frontiers in Genetics</i> , 2015, 6, 102.	1.1	13
282	POLG2 deficiency causes adult-onset syndromic sensory neuropathy, ataxia and parkinsonism. <i>Annals of Clinical and Translational Neurology</i> , 2017, 4, 4-14.	1.7	13
283	Inherited pathogenic mitochondrial DNA mutations and gastrointestinal stem cell populations. <i>Journal of Pathology</i> , 2018, 246, 427-432.	2.1	13
284	A novel mitochondrial tRNAGlu (MTTE) gene mutation causing chronic progressive external ophthalmoplegia at low levels of heteroplasmy in muscle. <i>Journal of the Neurological Sciences</i> , 2010, 298, 140-144.	0.3	12
285	Peripheral neuropathy in patients with CPEO associated with single and multiple mtDNA deletions. <i>Neurology: Genetics</i> , 2016, 2, e113.	0.9	12
286	Novel compound mutations in the mitochondrial translation elongation factor (TSFM) gene cause severe cardiomyopathy with myocardial fibro-adipose replacement. <i>Scientific Reports</i> , 2019, 9, 5108.	1.6	12
287	Elucidating the molecular mechanisms associated with <i>TARS2</i> -related mitochondrial disease. <i>Human Molecular Genetics</i> , 2022, 31, 523-534.	1.4	12
288	False-Positive Diagnosis of a Single, Large-Scale Mitochondrial DNA Deletion by Southern Blot Analysis: The Role of Neutral Polymorphisms. <i>Genetic Testing and Molecular Biomarkers</i> , 2004, 8, 395-399.	1.7	11

#	ARTICLE	IF	CITATIONS
289	<i>OPA1</i> mutations induce mtDNA proliferation in leukocytes of patients with dominant optic atrophy. <i>Neurology</i> , 2012, 79, 1515-1517.	1.5	11
290	De novo <i>CTBP1</i> variant is associated with decreased mitochondrial respiratory chain activities. <i>Neurology: Genetics</i> , 2017, 3, e187.	0.9	11
291	A Novel Pathogenic Variant in MT-CO2 Causes an Isolated Mitochondrial Complex IV Deficiency and Late-Onset Cerebellar Ataxia. <i>Journal of Clinical Medicine</i> , 2019, 8, 789.	1.0	11
292	Lewy body pathology is more prevalent in older individuals with mitochondrial disease than controls. <i>Acta Neuropathologica</i> , 2020, 139, 219-221.	3.9	11
293	Diabetes and Deafness: Is it sufficient to screen for the mitochondrial 3243A>G mutation alone?. <i>Diabetes Care</i> , 2007, 30, 2238-2239.	4.3	10
294	Testing the adaptive selection of human mtDNA haplogroups: an experimental bioenergetics approach. <i>Biochemical Journal</i> , 2007, 404, e3-5.	1.7	10
295	Mutations in the mitochondrial tRNA ^{Ser} (AGY) gene are associated with deafness, retinal degeneration, myopathy and epilepsy. <i>European Journal of Human Genetics</i> , 2012, 20, 897-904.	1.4	10
296	A novel m.7539C>T point mutation in the mt-tRNA ^{Asp} gene associated with multisystemic mitochondrial disease. <i>Neuromuscular Disorders</i> , 2015, 25, 81-84.	0.3	10
297	Novel GFM2 variants associated with early-onset neurological presentations of mitochondrial disease and impaired expression of OXPHOS subunits. <i>Neurogenetics</i> , 2017, 18, 227-235.	0.7	10
298	Disclosing the functional changes of two genetic alterations in a patient with Chronic Progressive External Ophthalmoplegia: Report of the novel mtDNA m.7486G>A variant. <i>Neuromuscular Disorders</i> , 2018, 28, 350-360.	0.3	10
299	Cognitive deficits in adult m.3243A>G and m.8344A>G related mitochondrial disease: importance of correcting for baseline intellectual ability. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 826-836.	1.7	10
300	SURF1 related Leigh syndrome: Clinical and molecular findings of 16 patients from Turkey. <i>Molecular Genetics and Metabolism Reports</i> , 2020, 25, 100657.	0.4	10
301	Neuromuscular Junction Abnormalities in Mitochondrial Disease. <i>Neurology: Clinical Practice</i> , 2021, 11, 97-104.	0.8	10
302	Interrogating Mitochondrial Biology and Disease Using CRISPR/Cas9 Gene Editing. <i>Genes</i> , 2021, 12, 1604.	1.0	10
303	Gene therapy for the treatment of mitochondrial DNA disorders. <i>Expert Opinion on Biological Therapy</i> , 2005, 5, 183-194.	1.4	9
304	Mitochondrial DNA abnormalities in ophthalmological disease. <i>Saudi Journal of Ophthalmology</i> , 2011, 25, 395-404.	0.3	9
305	A novel mitochondrial DNA m.7507A>G mutation is only pathogenic at high levels of heteroplasmy. <i>Neuromuscular Disorders</i> , 2015, 25, 262-267.	0.3	9
306	Rapid identification of human muscle disease with fibre optic Raman spectroscopy. <i>Analyst</i> , The, 2022, 147, 2533-2540.	1.7	9

#	ARTICLE	IF	CITATIONS
307	Toward a mtDNA locus-specific mutation database using the LOVD platform. <i>Human Mutation</i> , 2012, 33, 1352-1358.	1.1	8
308	Preliminary Evaluation of Clinician Rated Outcome Measures in Mitochondrial Disease. <i>Journal of Neuromuscular Diseases</i> , 2015, 2, 151-155.	1.1	8
309	Pigmentary retinopathy, rod cone dysfunction and sensorineural deafness associated with a rare mitochondrial tRNA ^{Lys} (m.8340G>A) gene variant. <i>British Journal of Ophthalmology</i> , 2017, 101, 1298-1302.	2.1	8
310	NEW OBSERVATIONS REGARDING THE RETINOPATHY OF GENETICALLY CONFIRMED KEARNS SAYRE SYNDROME. <i>Retinal Cases and Brief Reports</i> , 2018, 12, 349-358.	0.3	8
311	A novel, pathogenic dinucleotide deletion in the mitochondrial MT-TY gene causing myasthenia-like features. <i>Neuromuscular Disorders</i> , 2020, 30, 661-668.	0.3	8
312	Early onset coenzyme Q10 deficiency associated with ataxia and respiratory chain dysfunction due to novel pathogenic <i>COQ8A</i> variants, including a large intragenic deletion. <i>JIMD Reports</i> , 2020, 54, 45-53.	0.7	8
313	The Effect of tRNA ^{[Ser]^{Sec}} Isopentenylolation on Selenoprotein Expression. <i>International Journal of Molecular Sciences</i> , 2021, 22, 11454.	1.8	8
314	The pathogenic m.3243A>T mitochondrial DNA mutation is associated with a variable neurological phenotype. <i>Neuromuscular Disorders</i> , 2010, 20, 403-406.	0.3	7
315	The m.3291T>C mt-tRNA ^{Leu} (UUR) mutation is definitely pathogenic and causes multisystem mitochondrial disease. <i>Journal of the Neurological Sciences</i> , 2013, 325, 165-169.	0.3	7
316	Identification of a novel heterozygous guanosine monophosphate reductase (<i>GMPR</i>) variant in a patient with a late onset disorder of mitochondrial DNA maintenance. <i>Clinical Genetics</i> , 2020, 97, 276-286.	1.0	7
317	Neuromuscular disease presentation with three genetic defects involving two genomes. <i>Neuromuscular Disorders</i> , 2009, 19, 841-844.	0.3	6
318	Diagnostic investigations of patients with chronic progressive external ophthalmoplegia. <i>British Journal of Ophthalmology</i> , 2012, 96, 1536.2-1536.	2.1	6
319	Periventricular Calcification, Abnormal Pterins and Dry Thickened Skin: Expanding the Clinical Spectrum of RMND1?. <i>JIMD Reports</i> , 2015, 26, 13-19.	0.7	6
320	Clinical features of the pathogenic m.5540G>A mitochondrial transfer RNA tryptophan gene mutation. <i>Neuromuscular Disorders</i> , 2016, 26, 702-705.	0.3	6
321	<i>SLC25A42</i> associated mitochondrial encephalomyopathy: Report of additional founder cases and functional characterization of a novel deletion. <i>JIMD Reports</i> , 2021, 60, 75-87.	0.7	6
322	Machine learning algorithms reveal the secrets of mitochondrial dynamics. <i>EMBO Molecular Medicine</i> , 2021, 13, e14316.	3.3	6
323	Detection of Mitochondrial DNA Variation in Human Cells. <i>Methods in Molecular Biology</i> , 2010, 628, 227-257.	0.4	6
324	An unusual case of congenital muscular dystrophy with normal serum CK level, external ophthalmoplegia, and white matter changes on brain MRI. <i>European Journal of Paediatric Neurology</i> , 2007, 11, 381-384.	0.7	5

#	ARTICLE	IF	CITATIONS
325	The swinging pendulum of biomarkers in mitochondrial disease. <i>Neurology</i> , 2016, 87, 2286-2287.	1.5	5
326	A novel MT-CO2 variant causing cerebellar ataxia and neuropathy: The role of muscle biopsy in diagnosis and defining pathogenicity. <i>Neuromuscular Disorders</i> , 2021, 31, 1186-1193.	0.3	5
327	Characterising a homozygous two-exon deletion in <i>UQCRC1</i> : comparing human and mouse phenotypes. <i>EMBO Molecular Medicine</i> , 2021, 13, e14397.	3.3	5
328	The application of Raman spectroscopy to the diagnosis of mitochondrial muscle disease: A preliminary comparison between fibre optic probe and microscope formats. <i>Journal of Raman Spectroscopy</i> , 2022, 53, 172-181.	1.2	5
329	Bridging PNAs can bind preferentially to a deleted mitochondrial DNA template but replication by mitochondrial DNA polymerase γ in vitro is not impaired. <i>Biochimica Et Biophysica Acta Gene Regulatory Mechanisms</i> , 2003, 1629, 73-83.	2.4	4
330	Phenotypic diversity associated with the mitochondrial m.8313G>A point mutation. <i>Muscle and Nerve</i> , 2009, 40, 648-651.	1.0	4
331	Insights into N-calls of mitochondrial DNA sequencing using MitoChip v2.0. <i>BMC Research Notes</i> , 2011, 4, 426.	0.6	4
332	A novel pathogenic m.4412G>A MT-TM mitochondrial DNA variant associated with childhood-onset seizures, myopathy and bilateral basal ganglia changes. <i>Mitochondrion</i> , 2019, 47, 18-23.	1.6	4
333	Progressive external ophthalmoplegia due to a recurrent de novo m.15990C>T MT-TP (mt-tRNAPro) gene variant. <i>Neuromuscular Disorders</i> , 2020, 30, 346-350.	0.3	4
334	Pathogenic SLC25A26 variants impair SAH transport activity causing mitochondrial disease. <i>Human Molecular Genetics</i> , 2022, 31, 2049-2062.	1.4	3
335	Novel <i>POLG</i> variants associated with late-onset de novo status epilepticus and progressive ataxia. <i>Neurology: Genetics</i> , 2017, 3, e181.	0.9	2
336	A novel mitochondrial m.4414T>C MT-TM gene variant causing progressive external ophthalmoplegia and myopathy. <i>Neuromuscular Disorders</i> , 2019, 29, 693-697.	0.3	2
337	Multisystem mitochondrial disease caused by a rare m.10038G>A mitochondrial tRNA ^{Gly} (<i>MT-TG</i>) variant. <i>Neurology: Genetics</i> , 2020, 6, e413.	0.9	2
338	Chronic Progressive External Ophthalmoplegia due to a Rare de novo m.12334G>A MT-TL2 Mitochondrial DNA Variant1. <i>Journal of Neuromuscular Diseases</i> , 2020, 7, 355-360.	1.1	2
339	Chapter 7 Current and Future Prospects for the Treatment of Mitochondrial Disorders. <i>Blue Books of Practical Neurology</i> , 2002, 26, 213-227.	0.1	1
340	Mitochondrial respiratory chain defects and developmental diaphragmatic dysfunction in the neonatal period. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2006, 19, 587-589.	0.7	1
341	Modelling mitochondrial DNA mutations in bacterial cytochrome <i>c</i> oxidase: Link to colon cancer?. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2009, 106, E57.	3.3	1
342	Diabetes Mellitus in Mitochondrial Disease. <i>Frontiers in Diabetes</i> , 2017, , 55-68.	0.4	1

#	ARTICLE	IF	CITATIONS
343	Opening One's Eyes to Mosaicism in Progressive External Ophthalmoplegia. <i>Neurology: Genetics</i> , 2017, 3, e202.	0.9	1
344	Laboratory Investigation of Mitochondrial Diseases. , 0, , 39-51.		1
345	Uniparental isodisomy of chromosome 2 causing MRPL44-related multisystem mitochondrial disease. <i>Molecular Biology Reports</i> , 2021, 48, 2093-2104.	1.0	1
346	Novel phosphopantothienoylcysteine synthetase (<i>PPCS</i>) mutations with prominent neuromuscular features: Expanding the phenotypical spectrum of <i>PPCS</i> -related disorders. <i>American Journal of Medical Genetics, Part A</i> , 0, , .	0.7	1
347	Investigation of the mitochondrial genome in patients with atypical motor neuron disease. <i>Journal of Neurology</i> , 2007, 254, 482-487.	1.8	0
348	Nuclear genetic disorders of mitochondrial DNA gene expression. , 2020, , 375-409.		0
349	mtDNA disease for the neurologist. <i>Future Neurology</i> , 2011, 6, 63-80.	0.9	0
350	Selective Inhibition of Mutant Mitochondrial DNA Replication. <i>Expert Opinion on Therapeutic Targets</i> , 1997, 1, 249-252.	1.0	0
351	The Use of PNAs and Their Derivatives in Mitochondrial Gene Therapy. , 2006, , 243-251.		0