Robert W Taylor

List of Publications by Year
in descending order

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		6254	8630
351	27,261	80	146
papers	citations	h-index	g-index

356	356	356	22491
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mitochondrial DNA mutations in human disease. Nature Reviews Genetics, 2005, 6, 389-402.	16.3	1,530
2	High levels of mitochondrial DNA deletions in substantia nigra neurons in aging and Parkinson disease. Nature Genetics, 2006, 38, 515-517.	21.4	1,363
3	Prevalence of nuclear and mitochondrial <scp>DNA</scp> mutations related to adult mitochondrial disease. Annals of Neurology, 2015, 77, 753-759.	5.3	706
4	Prevalence of mitochondrial DNA disease in adults. Annals of Neurology, 2008, 63, 35-39.	5.3	540
5	Mitochondrial DNA mutations and human disease. Biochimica Et Biophysica Acta - Bioenergetics, 2010, 1797, 113-128.	1.0	527
6	Mitochondrial DNA mutations in human colonic crypt stem cells. Journal of Clinical Investigation, 2003, 112, 1351-1360.	8.2	454
7	Genetic diagnosis of Mendelian disorders via RNA sequencing. Nature Communications, 2017, 8, 15824.	12.8	432
8	Pronuclear transfer in human embryos to prevent transmission of mitochondrial DNA disease. Nature, 2010, 465, 82-85.	27.8	421
9	Phenotypic spectrum associated with mutations of the mitochondrial polymerase Â gene. Brain, 2006, 129, 1674-1684.	7.6	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. Brain, 2008, 131, 329-337.	7.6	381
11	Universal heteroplasmy of human mitochondrial DNA. Human Molecular Genetics, 2013, 22, 384-390.	2.9	344
12	What causes mitochondrial DNA deletions in human cells?. Nature Genetics, 2008, 40, 275-279.	21.4	334
13	The genetics and pathology of mitochondrial disease. Journal of Pathology, 2017, 241, 236-250.	4.5	329
14	The epidemiology of mitochondrial disordersâ€”past, present and future. Biochimica Et Biophysica Acta - Bioenergetics, 2004, 1659, 115-120.	1.0	325
15	Biochemical Assays of Respiratory Chain Complex Activity. Methods in Cell Biology, 2007, 80, 93-119.	1.1	324
16	A neurological perspective on mitochondrial disease. Lancet Neurology, The, 2010, 9, 829-840.	10.2	316
17	Use of Whole-Exome Sequencing to Determine the Genetic Basis of Multiple Mitochondrial Respiratory Chain Complex Deficiencies. JAMA - Journal of the American Medical Association, 2014, 312, 68.	7.4	304
18	Detection and quantification of mitochondrial DNA deletions in individual cells by real-time PCR. Nucleic Acids Research, 2002, 30, 68e-68.	14.5	279

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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.	7.1	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.	6.2	263
21	Selective inhibition of mutant human mitochondrial DNA replication in vitro by peptide nucleic acids. Nature Genetics, 1997, 15, 212-215.	21.4	252
22	Mutations causing mitochondrial disease: What is new and what challenges remain?. Science, 2015, 349, 1494-1499.	12.6	251
23	Mitochondrial dysfunction in a cell culture model of familial amyotrophic lateral sclerosis. Brain, 2002, 125, 1522-1533.	7.6	249
24	Mitochondrial Protein Interaction Mapping Identifies Regulators of Respiratory Chain Function. Molecular Cell, 2016, 63, 621-632.	9.7	241
25	Mitochondrial DNA and disease. Journal of Pathology, 2012, 226, 274-286.	4.5	239
26	Mechanisms of Field Cancerization in the Human Stomach: The Expansion and Spread of Mutated Gastric Stem Cells. Gastroenterology, 2008, 134, 500-510.	1.3	222
27	Recent Advances in Mitochondrial Disease. Annual Review of Genomics and Human Genetics, 2017, 18, 257-275.	6.2	217
28	Mutant POLG2 Disrupts DNA Polymerase Î³ Subunits and Causes Progressive External Ophthalmoplegia. American Journal of Human Genetics, 2006, 78, 1026-1034.	6.2	215
29	Superoxide dismutase downregulation in osteoarthritis progression and end-stage disease. Annals of the Rheumatic Diseases, 2010, 69, 1502-1510.	0.9	202
30	Risk of developing a mitochondrial DNA deletion disorder. Lancet, The, 2004, 364, 592-596.	13.7	201
31	mt <scp>DNA</scp> heteroplasmy level and copy number indicate disease burden in m.3243A>G mitochondrial disease. EMBO Molecular Medicine, 2018, 10, .	6.9	199
32	The UK MRC Mitochondrial Disease Patient Cohort Study: clinical phenotypes associated with the m.3243A>G mutation–implications for diagnosis and management. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 936-938.	1.9	193
33	Endurance training and detraining in mitochondrial myopathies due to single large-scale mtDNA deletions. Brain, 2006, 129, 3391-3401.	7.6	189
34	Cardiac involvement in mitochondrial DNA disease: clinical spectrum, diagnosis, and management. European Heart Journal, 2012, 33, 3023-3033.	2.2	182
35	The diagnosis of mitochondrial muscle disease. Neuromuscular Disorders, 2004, 14, 237-245.	0.6	178
36	The Spectrum of Mitochondrial Ultrastructural Defects in Mitochondrial Myopathy. Scientific Reports, 2016, 6, 30610.	3.3	165

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37	De novo mutations in the mitochondrialND3 gene as a cause of infantile mitochondrial encephalopathy and complex I deficiency. Annals of Neurology, 2004, 55, 58-64.	5.3	164
38	Induction of Mitochondrial Biogenesis Is a Maladaptive Mechanism in Mitochondrial Cardiomyopathies. Journal of the American College of Cardiology, 2007, 50, 1362-1369.	2.8	164
39	NDUFS6 mutations are a novel cause of lethal neonatal mitochondrial complex I deficiency. Journal of Clinical Investigation, 2004, 114, 837-845.	8.2	164
40	Multiple neonatal deaths due to a homoplasmic mitochondrial DNA mutation. Nature Genetics, 2002, 30, 145-146.	21.4	162
41	A homoplasmic mitochondrial transfer Ribonucleic Acid mutation as a cause of maternally inherited hypertrophic cardiomyopathy. Journal of the American College of Cardiology, 2003, 41, 1786-1796.	2.8	161
42	Assigning pathogenicity to mitochondrial tRNA mutations: when â€˜definitely maybeâ€™ is not good enough. Trends in Genetics, 2004, 20, 591-596.	6.7	159
43	A comparative analysis approach to determining the pathogenicity of mitochondrial tRNA mutations. Human Mutation, 2011, 32, 1319-1325.	2.5	159
44	Dominant inheritance of premature ovarian failure associated with mutant mitochondrial DNA polymerase gamma. Human Reproduction, 2006, 21, 2467-2473.	0.9	153
45	Mutations in the SPG7 gene cause chronic progressive external ophthalmoplegia through disordered mitochondrial DNA maintenance. Brain, 2014, 137, 1323-1336.	7.6	151
46	Resistance training in patients with single, large-scale deletions of mitochondrial DNA. Brain, 2008, 131, 2832-2840.	7.6	147
47	Mitochondrial tRNA mutations and disease. Wiley Interdisciplinary Reviews RNA, 2010, 1, 304-324.	6.4	145
48	Mutations in FBXL4 Cause Mitochondrial Encephalopathy and a Disorder of Mitochondrial DNA Maintenance. American Journal of Human Genetics, 2013, 93, 471-481.	6.2	137
49	Locating the stem cell niche and tracing hepatocyte lineages in human liver. Hepatology, 2009, 49, 1655-1663.	7.3	135
50	ELAC2 Mutations Cause a Mitochondrial RNA Processing Defect Associated with Hypertrophic Cardiomyopathy. American Journal of Human Genetics, 2013, 93, 211-223.	6.2	127
51	Late-onset optic atrophy, ataxia, and myopathy associated with a mutation of a complex II gene. Annals of Neurology, 2000, 48, 330-335.	5.3	126
52	Mutations in GTPBP3 Cause a Mitochondrial Translation Defect Associated with Hypertrophic Cardiomyopathy, Lactic Acidosis, and Encephalopathy. American Journal of Human Genetics, 2014, 95, 708-720.	6.2	123
53	The GABA Transaminase, ABAT, Is Essential for Mitochondrial Nucleoside Metabolism. Cell Metabolism, 2015, 21, 417-427.	16.2	119
54	Quantitative 3D Mapping of the Human Skeletal Muscle Mitochondrial Network. Cell Reports, 2019, 26, 996-1009.e4.	6.4	116

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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. American Journal of Human Genetics, 1999, 64, 1330-1339.	6.2	115
56	The neurology of mitochondrial DNA disease. Lancet Neurology, The, 2002, 1, 343-351.	10.2	115
57	Clonal Expansion of Early to Mid-Life Mitochondrial DNA Point Mutations Drives Mitochondrial Dysfunction during Human Ageing. PLoS Genetics, 2014, 10, e1004620.	3.5	115
58	Mitochondrial dysfunction in osteoarthritis is associated with downâ€regulation of superoxide dismutase 2. Arthritis and Rheumatism, 2013, 65, 378-387.	6.7	113
59	SANDO syndrome in a cohort of 107 patients with CPEO and mitochondrial DNA deletions. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 630-634.	1.9	113
60	Recent advances in understanding the molecular genetic basis of mitochondrial disease. Journal of Inherited Metabolic Disease, 2020, 43, 36-50.	3.6	113
61	Molecular basis of infantile reversible cytochrome c oxidase deficiency myopathy. Brain, 2009, 132, 3165-3174.	7.6	112
62	Defective i6A37 Modification of Mitochondrial and Cytosolic tRNAs Results from Pathogenic Mutations in TRIT1 and Its Substrate tRNA. PLoS Genetics, 2014, 10, e1004424.	3.5	112
63	Biallelic Mutations in NBAS Cause Recurrent Acute Liver Failure with Onset in Infancy. American Journal of Human Genetics, 2015, 97, 163-169.	6.2	110
64	Noninvasive diagnosis of the 3243A>G mitochondrial DNA mutation using urinary epithelial cells. European Journal of Human Genetics, 2004, 12, 778-781.	2.8	107
65	Cell-permeable succinate prodrugs bypass mitochondrial complex I deficiency. Nature Communications, 2016, 7, 12317.	12.8	106
66	A novel immunofluorescent assay to investigate oxidative phosphorylation deficiency in mitochondrial myopathy: understanding mechanisms and improving diagnosis. Scientific Reports, 2015, 5, 15037.	3.3	104
67	Disease progression in patients with single, large-scale mitochondrial DNA deletions. Brain, 2014, 137, 323-334.	7.6	103
68	Respiratory chain complex I deficiency caused by mitochondrial DNA mutations. European Journal of Human Genetics, 2011, 19, 769-775.	2.8	102
69	Topoisomerase 3Î± Is Required for Decatenation and Segregation of Human mtDNA. Molecular Cell, 2018, 69, 9-23.e6.	9.7	102
70	Phenotypic heterogeneity in m.3243A>G mitochondrial disease: The role of nuclear factors. Annals of Clinical and Translational Neurology, 2018, 5, 333-345.	3.7	102
71	Recessive germlineSDHAandSDHBmutations causing leukodystrophy and isolated mitochondrial complex II deficiency. Journal of Medical Genetics, 2012, 49, 569-577.	3.2	100
72	Mitochondrial disease in adults: recent advances and future promise. Lancet Neurology, The, 2021, 20, 573-584.	10.2	96

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73	Endocrine disorders in mitochondrial disease. Molecular and Cellular Endocrinology, 2013, 379, 2-11.	3.2	93
74	Recurrent De Novo Dominant Mutations in SLC25A4 Cause Severe Early-Onset Mitochondrial Disease and Loss of Mitochondrial DNA Copy Number. American Journal of Human Genetics, 2016, 99, 860-876.	6.2	93
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91	Succinateâ€CoA ligase deficiency due to mutations in SUCLA2 and SUCLG1: phenotype and genotype correlations in 71 patients. Journal of Inherited Metabolic Disease, 2016, 39, 243-252.	3.6	79
92	A multiplex real-time PCR method to detect and quantify mitochondrial DNA deletions in individual cells. Analytical Biochemistry, 2007, 370, 127-129.	2.4	77
93	Impaired mitochondrial biogenesis is a common feature to myocardial hypertrophy and end-stage ischemic heart failure. Cardiovascular Pathology, 2016, 25, 103-112.	1.6	77
94	Genotypes from patients indicate no paternal mitochondrial DNA contribution. Annals of Neurology, 2003, 54, 521-524.	5.3	76
95	Overexpression of human mitochondrial valyl tRNA synthetase can partially restore levels of cognate mt-tRNAVal carrying the pathogenic C25U mutation. Nucleic Acids Research, 2008, 36, 3065-3074.	14.5	74
96	Mitochondrial OXPHOS Biogenesis: Co-Regulation of Protein Synthesis, Import, and Assembly Pathways. International Journal of Molecular Sciences, 2020, 21, 3820.	4.1	74
97	Mitochondrial and inflammatory changes in sporadic inclusion body myositis. Neuropathology and Applied Neurobiology, 2015, 41, 288-303.	3.2	73
98	Retrospective natural history of thymidine kinase 2 deficiency. Journal of Medical Genetics, 2018, 55, 515-521.	3.2	73
99	Depletion of mitochondrial DNA in leucocytes harbouring the 3243A->G mtDNA mutation. Journal of Medical Genetics, 2006, 44, 69-74.	3.2	72
100	A Methodological Approach to Tracing Cell Lineage in Human Epithelial Tissues. Stem Cells, 2009, 27, 1410-1420.	3.2	72
101	Familial myopathy: New insights into the T14709C mitochondrial tRNA mutation. Annals of Neurology, 2004, 55, 478-484.	5.3	71
102	Adults with RRM2B-related mitochondrial disease have distinct clinical and molecular characteristics. Brain, 2012, 135, 3392-3403.	7.6	70
103	Deficiency of complex II of the mitochondrial respiratory chain in late-onset optic atrophy and ataxia. Annals of Neurology, 1996, 39, 224-232.	5.3	69
104	The p.M292T NDUFS2 mutation causes complex I-deficient Leigh syndrome in multiple families. Brain, 2010, 133, 2952-2963.	7.6	69
105	Clinical, Genetic, and Radiological Features of Extrapyramidal Movement Disorders in Mitochondrial Disease. JAMA Neurology, 2016, 73, 668.	9.0	69
106	Age-associated mitochondrial DNA mutations cause metabolic remodeling that contributes to accelerated intestinal tumorigenesis. Nature Cancer, 2020, 1, 976-989.	13.2	69
107	Isoleucyl-tRNA synthetase levels modulate the penetrance of a homoplasmic m.4277T>C mitochondrial tRNAIle mutation causing hypertrophic cardiomyopathy. Human Molecular Genetics, 2012, 21, 85-100.	2.9	67
108	MTO1 Mutations are Associated with Hypertrophic Cardiomyopathy and Lactic Acidosis and Cause Respiratory Chain Deficiency in Humans and Yeast. Human Mutation, 2013, 34, 1501-1509.	2.5	67

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109	MPV17 Loss Causes Deoxynucleotide Insufficiency and Slow DNA Replication in Mitochondria. PLoS Genetics, 2016, 12, e1005779.	3.5	67
110	Nuclear factors involved in mitochondrial translation cause a subgroup of combined respiratory chain deficiency. Brain, 2011, 134, 183-195.	7.6	66
111	Accurate mitochondrial DNA sequencing using off-target reads provides a single test to identify pathogenic point mutations. Genetics in Medicine, 2014, 16, 962-971.	2.4	64
112	Mutations in APOPT1, Encoding a Mitochondrial Protein, Cause Cavitating Leukoencephalopathy with Cytochrome c Oxidase Deficiency. American Journal of Human Genetics, 2014, 95, 315-325.	6.2	64
113	Multipotent Basal Stem Cells, Maintained in Localized Proximal Niches, Support Directed Long-Ranging Epithelial Flows in Human Prostates. Cell Reports, 2017, 20, 1609-1622.	6.4	64
114	Mutations in MDH2, Encoding a Krebs Cycle Enzyme, Cause Early-Onset Severe Encephalopathy. American Journal of Human Genetics, 2017, 100, 151-159.	6.2	63
115	In situ lineage tracking of human prostatic epithelial stem cell fate reveals a common clonal origin for basal and luminal cells. Journal of Pathology, 2011, 225, 181-188.	4.5	62
116	Pathogenic Mitochondrial t <scp>RNA</scp> Point Mutations: Nine Novel Mutations Affirm Their Importance as a Cause of Mitochondrial Disease. Human Mutation, 2013, 34, 1260-1268.	2.5	62
117	Epilepsy in adults with mitochondrial disease: A cohort study. Annals of Neurology, 2015, 78, 949-957.	5.3	62
118	A novel ANT1 gene mutation with probable germline mosaicism in autosomal dominant progressive external ophthalmoplegia. Neuromuscular Disorders, 2005, 15, 311-315.	0.6	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. PLoS Genetics, 2012, 8, e1003082.	3.5	61
120	Mutation of the human mitochondrial phenylalanine-tRNA synthetase causes infantile-onset epilepsy and cytochrome c oxidase deficiency. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2014, 1842, 56-64.	3.8	61
121	Sensory neuronopathy in patients harbouring recessive polymerase Î³ mutations. Brain, 2012, 135, 62-71.	7.6	60
122	Treatment of mitochondrial disease. Journal of Bioenergetics and Biomembranes, 1997, 29, 195-205.	2.3	59
123	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. American Journal of Human Genetics, 2018, 102, 494-504.	6.2	59
124	Targeting large molecules to mitochondria. Advanced Drug Delivery Reviews, 2001, 49, 189-198.	13.7	58
125	A novel sporadic mutation in cytochrome c oxidase subunit II as a cause of rhabdomyolysis. Neuromuscular Disorders, 2004, 14, 162-166.	0.6	58
126	Mitochondrial DNA Defects and Selective Extraocular Muscle Involvement in CPEO. , 2010, 51, 3340.		58

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127	Biallelic C1QBP Mutations Cause Severe Neonatal-, Childhood-, or Later-Onset Cardiomyopathy Associated with Combined Respiratory-Chain Deficiencies. American Journal of Human Genetics, 2017, 101, 525-538.	6.2	58
128	Neonatal Fanconi syndrome due to deficiency of complex III of the respiratory chain. Pediatric Nephrology, 1995, 9, 407-411.	1.7	57
129	A national perspective on prenatal testing for mitochondrial disease. European Journal of Human Genetics, 2014, 22, 1255-1259.	2.8	57
130	Biallelic Mutations in TMEM126B Cause Severe Complex I Deficiency with a Variable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 217-227.	6.2	57
131	Succinate-cytochrome c reductase: assessment of its value in the investigation of defects of the respiratory chain. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 1993, 1181, 261-265.	3.8	56
132	Ageâ€associated mitochondrial DNA mutations lead to small but significant changes in cell proliferation and apoptosis in human colonic crypts. Aging Cell, 2010, 9, 96-99.	6.7	56
133	Nucleotide pools dictate the identity and frequency of ribonucleotide incorporation in mitochondrial DNA. PLoS Genetics, 2017, 13, e1006628.	3.5	55
134	Motor neuron disease in a patient with a mitochondrial tRNAIlemutation. Annals of Neurology, 2006, 59, 570-574.	5.3	54
135	De novo mtDNA point mutations are common and have a low recurrence risk. Journal of Medical Genetics, 2017, 54, 73-83.	3.2	54
136	<scp>OXA</scp> 1L mutations cause mitochondrial encephalopathy and a combined oxidative phosphorylation defect. EMBO Molecular Medicine, 2018, 10, .	6.9	54
137	Mitochondrial DNA sequence characteristics modulate the size of the genetic bottleneck. Human Molecular Genetics, 2016, 25, 1031-1041.	2.9	53
138	Sudden adult death syndrome in m.3243A>G-related mitochondrial disease: an unrecognized clinical entity in young, asymptomatic adults. European Heart Journal, 2016, 37, 2552-2559.	2.2	53
139	Accurate Measurement of Mitochondrial DNA Deletion Level and Copy Number Differences in Human Skeletal Muscle. PLoS ONE, 2014, 9, e114462.	2.5	53
140	Defects in multiple complexes of the respiratory chain are present in ageing human colonic crypts. Experimental Gerontology, 2010, 45, 573-579.	2.8	52
141	Clinical, biochemical, cellular and molecular characterization of mitochondrial DNA depletion syndrome due to novel mutations in the MPV17 gene. European Journal of Human Genetics, 2014, 22, 184-191.	2.8	52
142	MPV17 mutation causes neuropathy and leukoencephalopathy with multiple mtDNA deletions in muscle. Neuromuscular Disorders, 2012, 22, 587-591.	0.6	51
143	NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. Brain, 2019, 142, 50-58.	7.6	51
144	SCYL1 variants cause a syndrome with lowÎ³-glutamyl-transferase cholestasis, acute liver failure, and neurodegeneration (CALFAN). Genetics in Medicine, 2018, 20, 1255-1265.	2.4	50

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145	Pathogenic mitochondrial tRNA mutations - Which mutations are inherited and why?. Human Mutation, 2009, 30, E984-E992.	2.5	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.	3.8	49
147	Defining mitochondrial protein functions through deep multiomic profiling. Nature, 2022, 606, 382-388.	27.8	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.	1.0	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.	2.9	48
151	Ultrasensitive deletion detection links mitochondrial DNA replication, disease, and aging. Genome Biology, 2020, 21, 248.	8.8	48
152	Subcellular origin of mitochondrial DNA deletions in human skeletal muscle. Annals of Neurology, 2018, 84, 289-301.	5.3	47
153	What is influencing the phenotype of the common homozygous polymerase-Î³ mutation p.Ala467Thr?. Brain, 2012, 135, 3614-3626.	7.6	46
154	Structural modeling of tissue-specific mitochondrial alanyl-tRNA synthetase (AARS2) defects predicts differential effects on aminoacylation. Frontiers in Genetics, 2015, 6, 21.	2.3	46
155	Clinical, morphological, biochemical, imaging and outcome parameters in 21 individuals with mitochondrial maintenance defect related to FBXL4 mutations. Journal of Inherited Metabolic Disease, 2015, 38, 905-914.	3.6	45
156	Metabolic effects of bezafibrate in mitochondrial disease. EMBO Molecular Medicine, 2020, 12, e11589.	6.9	45
157	Microangiopathy in the cerebellum of patients with mitochondrial DNA disease. Brain, 2012, 135, 1736-1750.	7.6	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.	1.8	44
159	<scp>FBXL</scp> 4 deficiency increases mitochondrial removal by autophagy. EMBO Molecular Medicine, 2020, 12, e11659.	6.9	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.	1.7	43
161	The isolated carboxyâ€terminal domain of human mitochondrial leucylâ€<scp>tRNA</scp> synthetase rescues the pathological phenotype of mitochondrial <scp>tRNA</scp> mutations in human cells. EMBO Molecular Medicine, 2014, 6, 169-182.	6.9	43
162	A novel inborn error of the coenzyme Q10 biosynthesis pathway: cerebellar ataxia and static encephalomyopathy due to COQ5 Câ€methyltransferase deficiency. Human Mutation, 2018, 39, 69-79.	2.5	43

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163	Clinical presentation and proteomic signature of patients with TANGO2 mutations. Journal of Inherited Metabolic Disease, 2020, 43, 297-308.	3.6	43
164	Developmental and Pathological Changes in the Human Cardiac Muscle Mitochondrial DNA Organization, Replication and Copy Number. PLoS ONE, 2010, 5, e10426.	2.5	43
165	Novel mutations in the TK2 gene associated with fatal mitochondrial DNA depletion myopathy. Neuromuscular Disorders, 2008, 18, 557-560.	0.6	42
166	POLG1 Mutations Manifesting as Autosomal Recessive Axonal Charcot-Marie-Tooth Disease. Archives of Neurology, 2008, 65, 133-6.	4.5	42
167	A New Mitochondrial Transfer RNAPro Gene Mutation Associated With Myoclonic Epilepsy With Ragged-Red Fibers and Other Neurological Features. Archives of Neurology, 2009, 66, 399-402.	4.5	42
168	Adult-onset spinocerebellar ataxia syndromes due toMTATP6mutations. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 883-886.	1.9	42
169	Pathological mechanisms underlying single largeâ€scale mitochondrial <scp>DNA</scp> deletions. Annals of Neurology, 2018, 83, 115-130.	5.3	42
170	A novel Twinkle gene mutation in autosomal dominant progressive external ophthalmoplegia. Neuromuscular Disorders, 2003, 13, 568-572.	0.6	41
171	Clonal Expansion of Secondary Mitochondrial DNA Deletions Associated With Spinocerebellar Ataxia Type 28. JAMA Neurology, 2015, 72, 106.	9.0	41
172	The urinary proteome and metabonome differ from normal in adults with mitochondrial disease. Kidney International, 2015, 87, 610-622.	5.2	41
173	Clinical Features, Molecular Heterogeneity, and Prognostic Implications in YARS2-Related Mitochondrial Myopathy. JAMA Neurology, 2017, 74, 686.	9.0	41
174	Bi-allelic Mutations in NDUFA6 Establish Its Role in Early-Onset Isolated Mitochondrial Complex I Deficiency. American Journal of Human Genetics, 2018, 103, 592-601.	6.2	41
175	Incidence of Primary Mitochondrial Disease in Children Younger Than 2 Years Presenting With Acute Liver Failure. Journal of Pediatric Gastroenterology and Nutrition, 2016, 63, 592-597.	1.8	40
176	Pseudoâ€obstruction, stroke, and mitochondrial dysfunction: A lethal combination. Annals of Neurology, 2016, 80, 686-692.	5.3	40
177	Clonally Expanded Mitochondrial DNA Mutations in Epileptic Individuals With Mutated DNA Polymerase Î³. Journal of Neuropathology and Experimental Neurology, 2008, 67, 857-866.	1.7	39
178	Cytochrome c oxidase-intermediate fibres: Importance in understanding the pathogenesis and treatment of mitochondrial myopathy. Neuromuscular Disorders, 2012, 22, 690-698.	0.6	39
179	Pathogenic Bi-allelic Mutations in NDUFAF8 Cause Leigh Syndrome with an Isolated Complex I Deficiency. American Journal of Human Genetics, 2020, 106, 92-101.	6.2	39
180	Mitochondrial DNA mutations in human disease. IUBMB Life, 2006, 58, 143-151.	3.4	37

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181	Mitochondrial pathology in progressive cerebellar ataxia. Cerebellum and Ataxias, 2015, 2, 16.	1.9	37
182	Complex mitochondrial DNA rearrangements in individual cells from patients with sporadic inclusion body myositis. Nucleic Acids Research, 2016, 44, 5313-5329.	14.5	37
183	Clinical, biochemical, and genetic features of four patients with shortâ€chain enoylâ€CoA hydratase (ECHS1) deficiency. American Journal of Medical Genetics, Part A, 2018, 176, 1115-1127.	1.2	36
184	The clinical spectrum of the m.10191T>C mutation in complex Iâ€deficient Leigh syndrome. Developmental Medicine and Child Neurology, 2012, 54, 500-506.	2.1	35
185	The role of the mitochondrial ribosome in human disease: searching for mutations in 12S mitochondrial rRNA with high disruptive potential. Human Molecular Genetics, 2014, 23, 949-967.	2.9	35
186	The clinical, biochemical and genetic features associated withRMND1-related mitochondrial disease. Journal of Medical Genetics, 2016, 53, 768-775.	3.2	35
187	A Wars2 Mutant Mouse Model Displays OXPHOS Deficiencies and Activation of Tissue-Specific Stress Response Pathways. Cell Reports, 2018, 25, 3315-3328.e6.	6.4	35
188	A novel mitochondrial MTND5 frameshift mutation causing isolated complex I deficiency, renal failure and myopathy. Neuromuscular Disorders, 2010, 20, 131-135.	0.6	34
189	Bmi1 enhances skeletal muscle regeneration through MT1-mediated oxidative stress protection in a mouse model of dystrophinopathy. Journal of Experimental Medicine, 2014, 211, 2617-2633.	8.5	34
190	Dissecting the mechanisms underlying the accumulation of mitochondrial DNA deletions in human skeletal muscle. Human Molecular Genetics, 2014, 23, 4612-4620.	2.9	34
191	Copy-choice recombination during mitochondrial L-strand synthesis causes DNA deletions. Nature Communications, 2019, 10, 759.	12.8	34
192	POLG mutations cause decreased mitochondrial DNA repopulation rates following induced depletion in human fibroblasts. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2011, 1812, 321-325.	3.8	33
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194	Pathogenic variants in MTâ€ATP6: A United Kingdomâ€“based mitochondrial disease cohort study. Annals of Neurology, 2019, 86, 310-315.	5.3	33
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