Robert William Taylor

List of Publications by Year
in descending order

241 papers	17,681 citations	20817 60 h-index	17592 121 g-index
241 all docs	241 docs citations	241 times ranked	18027 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	Mitochondrial DNA mutations in human colonic crypt stem cells. Journal of Clinical Investigation, 2003, 112, 1351-1360.	8.2	454
5	Genetic diagnosis of Mendelian disorders via RNA sequencing. Nature Communications, 2017, 8, 15824.	12.8	432
6	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
7	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care â€" Preliminary Report. New England Journal of Medicine, 2021, 385, 1868-1880.	27.0	352
8	What causes mitochondrial DNA deletions in human cells?. Nature Genetics, 2008, 40, 275-279.	21.4	334
9	The genetics and pathology of mitochondrial disease. Journal of Pathology, 2017, 241, 236-250.	4.5	329
10	Biochemical Assays of Respiratory Chain Complex Activity. Methods in Cell Biology, 2007, 80, 93-119.	1.1	324
11	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
12	Detection and quantification of mitochondrial DNA deletions in individual cells by real-time PCR. Nucleic Acids Research, 2002, 30, 68e-68.	14.5	279
13	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
14	Selective inhibition of mutant human mitochondrial DNA replication in vitro by peptide nucleic acids. Nature Genetics, 1997, 15, 212-215.	21.4	252
15	Mutations causing mitochondrial disease: What is new and what challenges remain?. Science, 2015, 349, 1494-1499.	12.6	251
16	Mitochondrial Protein Interaction Mapping Identifies Regulators of Respiratory Chain Function. Molecular Cell, 2016, 63, 621-632.	9.7	241
17	Mitochondrial DNA and disease. Journal of Pathology, 2012, 226, 274-286.	4.5	239
18	Recent Advances in Mitochondrial Disease. Annual Review of Genomics and Human Genetics, 2017, 18, 257-275.	6.2	217

#	Article	IF	Citations
19	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
20	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
21	The diagnosis of mitochondrial muscle disease. Neuromuscular Disorders, 2004, 14, 237-245.	0.6	178
22	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
23	A comparative analysis approach to determining the pathogenicity of mitochondrial tRNA mutations. Human Mutation, 2011, 32, 1319-1325.	2.5	159
24	Mutations in the SPG7 gene cause chronic progressive external ophthalmoplegia through disordered mitochondrial DNA maintenance. Brain, 2014, 137, 1323-1336.	7.6	151
25	Mitochondrial tRNA mutations and disease. Wiley Interdisciplinary Reviews RNA, 2010, 1, 304-324.	6.4	145
26	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
27	ELAC2 Mutations Cause a Mitochondrial RNA Processing Defect Associated with Hypertrophic Cardiomyopathy. American Journal of Human Genetics, 2013, 93, 211-223.	6.2	127
28	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
29	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
30	The GABA Transaminase, ABAT, Is Essential for Mitochondrial Nucleoside Metabolism. Cell Metabolism, 2015, 21, 417-427.	16.2	119
31	Quantitative 3D Mapping of the Human Skeletal Muscle Mitochondrial Network. Cell Reports, 2019, 26, 996-1009.e4.	6.4	116
32	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
33	Recent advances in understanding the molecular genetic basis of mitochondrial disease. Journal of Inherited Metabolic Disease, 2020, 43, 36-50.	3.6	113
34	Molecular basis of infantile reversible cytochrome c oxidase deficiency myopathy. Brain, 2009, 132, 3165-3174.	7.6	112
35	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
36	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

#	Article	IF	Citations
37	Cell-permeable succinate prodrugs bypass mitochondrial complex I deficiency. Nature Communications, 2016, 7, 12317.	12.8	106
38	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
39	Topoisomerase 3Î± Is Required for Decatenation and Segregation of Human mtDNA. Molecular Cell, 2018, 69, 9-23.e6.	9.7	102
40	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
41	Mitochondrial disease in adults: recent advances and future promise. Lancet Neurology, The, 2021, 20, 573-584.	10.2	96
42	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
43	Recurrent acute liver failure due to NBAS deficiency: phenotypic spectrum, disease mechanisms, and therapeutic concepts. Journal of Inherited Metabolic Disease, 2016, 39, 3-16.	3.6	92
44	Fatal infantile mitochondrial encephalomyopathy, hypertrophic cardiomyopathy and optic atrophy associated with a homozygousOPA1mutation. Journal of Medical Genetics, 2016, 53, 127-131.	3.2	91
45	Nonrandom tissue distribution of mutant mtDNA. American Journal of Medical Genetics Part A, 1999, 85, 498-501.	2.4	90
46	Progressive mitochondrial disease resulting from a novel missense mutation in the mitochondrial DNA ND3 gene. Annals of Neurology, 2001, 50, 104-107.	5.3	89
47	Recessive Mutations in TRMT10C Cause Defects in Mitochondrial RNA Processing and Multiple Respiratory Chain Deficiencies. American Journal of Human Genetics, 2016, 98, 993-1000.	6.2	89
48	Leigh disease associated with a novel mitochondrial DNA ND5 mutation. European Journal of Human Genetics, 2002, 10, 141-144.	2.8	88
49	Homozygous deletion in MICU1 presenting with fatigue and lethargy in childhood. Neurology: Genetics, 2016, 2, e59.	1.9	86
50	MR-1S Interacts with PET100 and PET117 in Module-Based Assembly of Human Cytochrome c Oxidase. Cell Reports, 2017, 18, 1727-1738.	6.4	86
51	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38.	8.2	85
52	Developmental Consequences of Defective ATG7-Mediated Autophagy in Humans. New England Journal of Medicine, 2021, 384, 2406-2417.	27.0	84
53	TRMT5 Mutations Cause a Defect in Post-transcriptional Modification of Mitochondrial tRNA Associated with Multiple Respiratory-Chain Deficiencies. American Journal of Human Genetics, 2015, 97, 319-328.	6.2	83
54	LRPPRCmutations cause early-onset multisystem mitochondrial disease outside of the French-Canadian population. Brain, 2015, 138, 3503-3519.	7.6	81

#	Article	IF	Citations
55	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
56	Genotypes from patients indicate no paternal mitochondrial DNA contribution. Annals of Neurology, 2003, 54, 521-524.	5.3	76
57	Mitochondrial OXPHOS Biogenesis: Co-Regulation of Protein Synthesis, Import, and Assembly Pathways. International Journal of Molecular Sciences, 2020, 21, 3820.	4.1	74
58	Retrospective natural history of thymidine kinase 2 deficiency. Journal of Medical Genetics, 2018, 55, 515-521.	3.2	73
59	Adults with RRM2B-related mitochondrial disease have distinct clinical and molecular characteristics. Brain, 2012, 135, 3392-3403.	7.6	70
60	Clinical, Genetic, and Radiological Features of Extrapyramidal Movement Disorders in Mitochondrial Disease. JAMA Neurology, 2016, 73, 668.	9.0	69
61	MPV17 Loss Causes Deoxynucleotide Insufficiency and Slow DNA Replication in Mitochondria. PLoS Genetics, 2016, 12, e1005779.	3.5	67
62	Thiamine deficiency in childhood with attention to genetic causes: Survival and outcome predictors. Annals of Neurology, 2017, 82, 317-330.	5.3	65
63	Mutations in TOP3A Cause a Bloom Syndrome-like Disorder. American Journal of Human Genetics, 2018, 103, 221-231.	6.2	65
64	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
65	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
66	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
67	Progressive deafnessâ€“dystonia due to SERAC1 mutations: A study of 67 cases. Annals of Neurology, 2017, 82, 1004-1015.	5.3	63
68	A Novel Mitochondrial DNA Point Mutation in the tRNAlleGene: Studies in a Patient Presenting with Chronic Progressive External Ophthalmoplegia and Multiple Sclerosis. Biochemical and Biophysical Research Communications, 1998, 243, 47-51.	2.1	62
69	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
70	Epilepsy in adults with mitochondrial disease: A cohort study. Annals of Neurology, 2015, 78, 949-957.	5.3	62
71	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
72	Emerging roles of ATG7 in human health and disease. EMBO Molecular Medicine, 2021, 13, e14824.	6.9	61

#	Article	IF	Citations
73	Sensory neuronopathy in patients harbouring recessive polymerase Î³ mutations. Brain, 2012, 135, 62-71.	7.6	60
74	Treatment of mitochondrial disease. Journal of Bioenergetics and Biomembranes, 1997, 29, 195-205.	2.3	59
75	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
76	Epilepsy due to mutations in the mitochondrial polymerase gamma (<scp>POLG</scp>) gene: A clinical and molecular genetic review. Epilepsia, 2016, 57, 1531-1545.	5.1	58
77	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
78	LONP1 and mtHSP70 cooperate to promote mitochondrial protein folding. Nature Communications, 2021, 12, 265.	12.8	58
79	A national perspective on prenatal testing for mitochondrial disease. European Journal of Human Genetics, 2014, 22, 1255-1259.	2.8	57
80	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
81	Nucleotide pools dictate the identity and frequency of ribonucleotide incorporation in mitochondrial DNA. PLoS Genetics, 2017, 13, e1006628.	3.5	55
82	De novo mtDNA point mutations are common and have a low recurrence risk. Journal of Medical Genetics, 2017, 54, 73-83.	3.2	54
83	<scp>OXA</scp> 1L mutations cause mitochondrial encephalopathy and a combined oxidative phosphorylation defect. EMBO Molecular Medicine, 2018, 10, .	6.9	54
84	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.	2.9	53
85	Mitochondrial DNA sequence characteristics modulate the size of the genetic bottleneck. Human Molecular Genetics, 2016, 25, 1031-1041.	2.9	53
86	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
87	Accurate Measurement of Mitochondrial DNA Deletion Level and Copy Number Differences in Human Skeletal Muscle. PLoS ONE, 2014, 9, e114462.	2.5	53
88	MPV17 mutation causes neuropathy and leukoencephalopathy with multiple mtDNA deletions in muscle. Neuromuscular Disorders, 2012, 22, 587-591.	0.6	51
89	NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. Brain, 2019, 142, 50-58.	7.6	51
90	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

#	Article	IF	Citations
91	Pathogenic mitochondrial tRNA mutations - Which mutations are inherited and why?. Human Mutation, 2009, 30, E984-E992.	2.5	49
92	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
93	Defining mitochondrial protein functions through deep multiomic profiling. Nature, 2022, 606, 382-388.	27.8	49
94	Sudden Cardiac Death Due to Deficiency of the Mitochondrial Inorganic Pyrophosphatase PPA2. American Journal of Human Genetics, 2016, 99, 674-682.	6.2	48
95	Ultrasensitive deletion detection links mitochondrial DNA replication, disease, and aging. Genome Biology, 2020, 21, 248.	8.8	48
96	Resveratrol attenuates oxidative stress in mitochondrial Complex I deficiency: Involvement of SIRT3. Free Radical Biology and Medicine, 2016, 96, 190-198.	2.9	47
97	MT-ND5 Mutation Exhibits Highly Variable Neurological Manifestations at Low Mutant Load. EBioMedicine, 2018, 30, 86-93.	6.1	47
98	Subcellular origin of mitochondrial DNA deletions in human skeletal muscle. Annals of Neurology, 2018, 84, 289-301.	5.3	47
99	Defective mitochondrial protease LonP1 can cause classical mitochondrial disease. Human Molecular Genetics, 2018, 27, 1743-1753.	2.9	46
100	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
101	Metabolic effects of bezafibrate in mitochondrial disease. EMBO Molecular Medicine, 2020, 12, e11589.	6.9	45
102	<scp>FBXL</scp> 4 deficiency increases mitochondrial removal by autophagy. EMBO Molecular Medicine, 2020, 12, e11659.	6.9	44
103	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
104	Clinical presentation and proteomic signature of patients with TANGO2 mutations. Journal of Inherited Metabolic Disease, 2020, 43, 297-308.	3.6	43
105	Pathological mechanisms underlying single largeâ€scale mitochondrial <scp>DNA</scp> deletions. Annals of Neurology, 2018, 83, 115-130.	5.3	42
106	A novel Twinkle gene mutation in autosomal dominant progressive external ophthalmoplegia. Neuromuscular Disorders, 2003, 13, 568-572.	0.6	41
107	Clinical Features, Molecular Heterogeneity, and Prognostic Implications in YARS2-Related Mitochondrial Myopathy. JAMA Neurology, 2017, 74, 686.	9.0	41
108	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

#	Article	IF	Citations
109	Pseudoâ€obstruction, stroke, and mitochondrial dysfunction: A lethal combination. Annals of Neurology, 2016, 80, 686-692.	5.3	40
110	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
111	Mitochondrial pathology in progressive cerebellar ataxia. Cerebellum and Ataxias, 2015, 2, 16.	1.9	37
112	Complex mitochondrial DNA rearrangements in individual cells from patients with sporadic inclusion body myositis. Nucleic Acids Research, 2016, 44, 5313-5329.	14.5	37
113	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
114	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
115	The clinical, biochemical and genetic features associated withRMND1-related mitochondrial disease. Journal of Medical Genetics, 2016, 53, 768-775.	3.2	35
116	The m.13051G>A mitochondrial DNA mutation results in variable neurology and activated mitophagy. Neurology, 2016, 86, 1921-1923.	1.1	35
117	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
118	Mosaic dysfunction of mitophagy in mitochondrial muscle disease. Cell Metabolism, 2022, 34, 197-208.e5.	16.2	35
119	Cardiomyopathy is common in patients with the mitochondrial DNA m.3243A>G mutation and correlates with mutation load. Neuromuscular Disorders, 2012, 22, 592-596.	0.6	34
120	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
121	Dissecting the mechanisms underlying the accumulation of mitochondrial DNA deletions in human skeletal muscle. Human Molecular Genetics, 2014, 23, 4612-4620.	2.9	34
122	Bi-allelic Mutations in Phe-tRNA Synthetase Associated with a Multi-system Pulmonary Disease Support Non-translational Function. American Journal of Human Genetics, 2018, 103, 100-114.	6.2	34
123	Copy-choice recombination during mitochondrial L-strand synthesis causes DNA deletions. Nature Communications, 2019, 10, 759.	12.8	34
124	Recurrent De Novo NAHR Reciprocal Duplications in the ATAD3 Gene Cluster Cause a Neurogenetic Trait with Perturbed Cholesterol and Mitochondrial Metabolism. American Journal of Human Genetics, 2020, 106, 272-279.	6.2	33
125	The genetics of mitochondrial disease: dissecting mitochondrial pathology using multiâ€omic pipelines. Journal of Pathology, 2021, 254, 430-442.	4.5	33
126	Mitochondrial dysfunction in myofibrillar myopathy. Neuromuscular Disorders, 2016, 26, 691-701.	0.6	32

#	Article	IF	Citations
127	Assessment of mitochondrial respiratory chain enzymes in cells and tissues. Methods in Cell Biology, 2020, 155, 121-156.	1.1	32
128	Neuropathologic Characterization of Pontocerebellar Hypoplasia Type 6 Associated With Cardiomyopathy and Hydrops Fetalis and Severe Multisystem Respiratory Chain Deficiency due to NovelRARS2Mutations. Journal of Neuropathology and Experimental Neurology, 2015, 74, 688-703.	1.7	31
129	A recurrent mitochondrial p.Trp22ArgNDUFB3variant causes a distinctive facial appearance, short stature and a mild biochemical and clinical phenotype. Journal of Medical Genetics, 2016, 53, 634-641.	3.2	31
130	Mitochondrial oxodicarboxylate carrier deficiency is associated with mitochondrial DNA depletion and spinal muscular atrophyâ€“like disease. Genetics in Medicine, 2018, 20, 1224-1235.	2.4	31
131	Mutations inELAC2associated with hypertrophic cardiomyopathy impair mitochondrial tRNA 3â€²â€end processing. Human Mutation, 2019, 40, 1731-1748.	2.5	31
132	Triplex real-time PCRâ€“an improved method to detect a wide spectrum of mitochondrial DNA deletions in single cells. Scientific Reports, 2015, 5, 9906.	3.3	30
133	Preferential amplification of a human mitochondrial DNA deletion in vitro and in vivo. Scientific Reports, 2018, 8, 1799.	3.3	30
134	Mutations of the mitochondrial carrier translocase channel subunit TIM22 cause early-onset mitochondrial myopathy. Human Molecular Genetics, 2018, 27, 4135-4144.	2.9	30
135	The presence of highly disruptive 16S rRNA mutations in clinical samples indicates a wider role for mutations of the mitochondrial ribosome in human disease. Mitochondrion, 2015, 25, 17-27.	3.4	29
136	The molecular pathology of pathogenic mitochondrial tRNA variants. FEBS Letters, 2021, 595, 1003-1024.	2.8	29
137	Dysferlin mutations and mitochondrial dysfunction. Neuromuscular Disorders, 2016, 26, 782-788.	0.6	28
138	Confirming TDP2 mutation in spinocerebellar ataxia autosomal recessive 23 (SCAR23). Neurology: Genetics, 2018, 4, e262.	1.9	27
139	Distal weakness with respiratory insufficiency caused by the m.8344A>G â€œMERRFâ€•mutation. Neuromuscular Disorders, 2014, 24, 533-536.	0.6	26
140	Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults. JAMA Neurology, 2018, 75, 105.	9.0	26
141	Mitochondrial stress response triggered by defects in protein synthesis quality control. Life Science Alliance, 2019, 2, e201800219.	2.8	26
142	Bi-allelic variants in the mitochondrial RNase P subunit PRORP cause mitochondrial tRNA processing defects and pleiotropic multisystem presentations. American Journal of Human Genetics, 2021, 108, 2195-2204.	6.2	26
143	Scientific and Ethical Issues in Mitochondrial Donation. New Bioethics, 2018, 24, 57-73.	1.1	25
144	Resolving complexity in mitochondrial disease: Towards precision medicine. Molecular Genetics and Metabolism, 2019, 128, 19-29.	1.1	25

#	Article	IF	Citations
145	ITPase deficiency causes a Martsolf-like syndrome with a lethal infantile dilated cardiomyopathy. PLoS Genetics, 2019, 15, e1007605.	3.5	25
146	Pathogenic mitochondrial mt-tRNAAla variants are uniquely associated with isolated myopathy. European Journal of Human Genetics, 2015, 23, 1735-1738.	2.8	24
147	Pathogenic mtDNA mutations causing mitochondrial myopathy: The need for muscle biopsy. Neurology: Genetics, 2016, 2, e82.	1.9	24
148	The genotypic and phenotypic spectrum of MTO1 deficiency. Molecular Genetics and Metabolism, 2018, 123, 28-42.	1.1	24
149	Investigating complex <scp>I</scp> deficiency in <scp>P</scp>urkinje cells and synapses in patients with mitochondrial disease. Neuropathology and Applied Neurobiology, 2016, 42, 477-492.	3.2	23
150	Lethal Neonatal LTBL Associated with Biallelic EARS2 Variants: Case Report and Review of the Reported Neuroradiological Features. JIMD Reports, 2016, 33, 61-68.	1.5	23
151	Pathogenic variants in HTRA2 cause an earlyâ€onset mitochondrial syndrome associated with 3â€methylglutaconic aciduria. Journal of Inherited Metabolic Disease, 2017, 40, 121-130.	3.6	23
152	Adult Onset Leigh Syndrome in the Intensive Care Setting: A Novel Presentation of a C12orf65 Related Mitochondrial Disease. Journal of Neuromuscular Diseases, 2015, 2, 409-419.	2.6	22
153	Three families with â€˜de novoâ€™ m.3243A>G mutation. BBA Clinical, 2016, 6, 19-24.	4.1	22
154	Clinical, biochemical, and genetic features associated with VARS2 -related mitochondrial disease. Human Mutation, 2018, 39, 563-578.	2.5	22
155	The genetic basis of isolated mitochondrial complex II deficiency. Molecular Genetics and Metabolism, 2020, 131, 53-65.	1.1	22
156	A p.R369G POLG2 mutation associated with adPEO and multiple mtDNA deletions causes decreased affinity between polymerase Î³ subunits. Mitochondrion, 2012, 12, 313-319.	3.4	21
157	Novel MTND1 mutations cause isolated exercise intolerance, complex I deficiency and increased assembly factor expression. Clinical Science, 2015, 128, 895-904.	4.3	21
158	Loss-of-function mutations inISCA2disrupt 4Fe-4S cluster machinery and cause a fatal leukodystrophy with hyperglycinemia and mtDNA depletion. Human Mutation, 2018, 39, 537-549.	2.5	21
159	Further delineation of the phenotypic spectrum ofÂ ISCA2 defect: A report of ten new cases. European Journal of Paediatric Neurology, 2018, 22, 46-55.	1.6	21
160	POLRMT mutations impair mitochondrial transcription causing neurological disease. Nature Communications, 2021, 12, 1135.	12.8	21
161	A novel mitochondrial DNA tRNAIle (A4267G) mutation in a sporadic patient with mitochondrial myopathy. Neuromuscular Disorders, 2002, 12, 659-664.	0.6	20
162	Using a quantitative quadruple immunofluorescent assay to diagnose isolated mitochondrial Complex I deficiency. Scientific Reports, 2017, 7, 15676.	3.3	20

#	Article	IF	Citations
163	Expanding the phenotype of de novo SLC25A4 -linked mitochondrial disease to include mild myopathy. Neurology: Genetics, 2018, 4, e256.	1.9	20
164	Dissecting the neuronal vulnerability underpinning Alpersâ€™ syndrome: a clinical and neuropathological study. Brain Pathology, 2019, 29, 97-113.	4.1	20
165	Congenital myasthenic syndrome with mild intellectual disability caused by a recurrent SLC25A1 variant. European Journal of Human Genetics, 2020, 28, 373-377.	2.8	20
166	Adult-onset Mendelian PEO Associated with Mitochondrial Disease. Journal of Neuromuscular Diseases, 2014, 1, 119-133.	2.6	19
167	A leaky splicing mutation in NFU1 is associated with a particular biochemical phenotype. Consequences for the diagnosis. Mitochondrion, 2016, 26, 72-80.	3.4	19
168	Histiocytoid cardiomyopathy and microphthalmia with linear skin defects syndrome: phenotypes linked by truncating variants in NDUFB11. Journal of Physical Education and Sports Management, 2017, 3, a001271.	1.2	19
169	Instability of the mitochondrial alanyl-tRNA synthetase underlies fatal infantile-onset cardiomyopathy. Human Molecular Genetics, 2019, 28, 258-268.	2.9	19
170	Molecular genetic investigations identify new clinical phenotypes associated with BCS1L-related mitochondrial disease. Human Molecular Genetics, 2019, 28, 3766-3776.	2.9	19
171	Height as a Clinical Biomarker of Disease Burden in Adult Mitochondrial Disease. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 2057-2066.	3.6	19
172	Decreased male reproductive success in association with mitochondrial dysfunction. European Journal of Human Genetics, 2017, 25, 1162-1164.	2.8	18
173	A novel histochemistry assay to assess and quantify focal cytochrome c oxidase deficiency. Journal of Pathology, 2018, 245, 311-323.	4.5	17
174	Leigh syndrome caused by mutations in <scp>MTFMT</scp> is associated with a better prognosis. Annals of Clinical and Translational Neurology, 2019, 6, 515-524.	3.7	17
175	Biâ€allelic pathogenic variants in NDUFC2 cause earlyâ€onset Leigh syndrome and stalled biogenesis of complex I. EMBO Molecular Medicine, 2020, 12, e12619.	6.9	17
176	Natural History of Leigh Syndrome: A Study of Disease Burden and Progression. Annals of Neurology, 2022, 91, 117-130.	5.3	17
177	Expanding the clinical phenotype of IARS2-related mitochondrial disease. BMC Medical Genetics, 2018, 19, 196.	2.1	16
178	Understanding mitochondrial DNA maintenance disorders at the single muscle fibre level. Nucleic Acids Research, 2019, 47, 7430-7443.	14.5	16
179	Compound heterozygous RMND1 gene variants associated with chronic kidney disease, dilated cardiomyopathy and neurological involvement: a case report. BMC Research Notes, 2016, 9, 325.	1.4	15
180	Both mitochondrial DNA and mitonuclear gene mutations cause hearing loss through cochlear dysfunction. Brain, 2016, 139, e33-e33.	7.6	15

#	Article	IF	Citations
181	Clinically proven mtDNA mutations are not common in those with chronic fatigue syndrome. BMC Medical Genetics, 2017, 18, 29.	2.1	15
182	Mitochondrial DNA Transcription: Regulating the Power Supply. Cell, 2007, 130, 211-213.	28.9	14
183	The frequency of the m.1555Aâ€‰›â€‰G (MTRNR1) variant in UK patients with suspected mitochondrial deafness. Hearing, Balance and Communication, 2016, 14, 101-102.	0.4	14
184	The natural history of infantile mitochondrial DNA depletion syndrome due to RRM2B deficiency. Genetics in Medicine, 2020, 22, 199-209.	2.4	14
185	Novel MT-ND Gene Variants Causing Adult-Onset Mitochondrial Disease and Isolated Complex I Deficiency. Frontiers in Genetics, 2020, 11, 24.	2.3	14
186	Inherited pathogenic mitochondrial DNA mutations and gastrointestinal stem cell populations. Journal of Pathology, 2018, 246, 427-432.	4.5	13
187	Peripheral neuropathy in patients with CPEO associated with single and multiple mtDNA deletions. Neurology: Genetics, 2016, 2, e113.	1.9	12
188	Novel compound mutations in the mitochondrial translation elongation factor (TSFM) gene cause severe cardiomyopathy with myocardial fibro-adipose replacement. Scientific Reports, 2019, 9, 5108.	3.3	12
189	2-Deoxy-D-glucose couples mitochondrial DNA replication with mitochondrial fitness and promotes the selection of wild-type over mutant mitochondrial DNA. Nature Communications, 2021, 12, 6997.	12.8	12
190	De novo CTBP1 variant is associated with decreased mitochondrial respiratory chain activities. Neurology: Genetics, 2017, 3, e187.	1.9	11
191	A Novel Pathogenic Variant in MT-CO2 Causes an Isolated Mitochondrial Complex IV Deficiency and Late-Onset Cerebellar Ataxia. Journal of Clinical Medicine, 2019, 8, 789.	2.4	11
192	A novel m.7539C>T point mutation in the mt-tRNAAsp gene associated with multisystemic mitochondrial disease. Neuromuscular Disorders, 2015, 25, 81-84.	0.6	10
193	Novel GFM2 variants associated with early-onset neurological presentations of mitochondrial disease and impaired expression of OXPHOS subunits. Neurogenetics, 2017, 18, 227-235.	1.4	10
194	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. Neuromuscular Disorders, 2018, 28, 350-360.	0.6	10
195	Skeletal muscle mitochondrial oxidative phosphorylation function in idiopathic pulmonary arterial hypertension: in vivo and in vitro study. Pulmonary Circulation, 2018, 8, 1-5.	1.7	10
196	SURF1 related Leigh syndrome: Clinical and molecular findings of 16 patients from Turkey. Molecular Genetics and Metabolism Reports, 2020, 25, 100657.	1.1	10
197	Mitochondrial DNA mutations induce mitochondrial biogenesis and increase the tumorigenic potential of Hodgkin and Reedâ€“Sternberg cells. Carcinogenesis, 2020, 41, 1735-1745.	2.8	10
198	Neuromuscular Junction Abnormalities in Mitochondrial Disease. Neurology: Clinical Practice, 2021, 11, 97-104.	1.6	10

#	Article	IF	Citations
199	Gene therapy for the treatment of mitochondrial DNA disorders. Expert Opinion on Biological Therapy, 2005, 5, 183-194.	3.1	9
200	A novel mitochondrial DNA m.7507A>G mutation is only pathogenic at high levels of heteroplasmy. Neuromuscular Disorders, 2015, 25, 262-267.	0.6	9
201	Adult-onset Mendelian PEO Associated with Mitochondrial Disease. Journal of Neuromuscular Diseases, 2014, 1, 119-133.	2.6	9
202	Rapid identification of human muscle disease with fibre optic Raman spectroscopy. Analyst, The, 2022, 147, 2533-2540.	3.5	9
203	Preliminary Evaluation of Clinician Rated Outcome Measures in Mitochondrial Disease. Journal of Neuromuscular Diseases, 2015, 2, 151-155.	2.6	8
204	Pigmentary retinopathy, rodâ€“cone dysfunction and sensorineural deafness associated with a rare mitochondrial tRNALys(m.8340G>A) gene variant. British Journal of Ophthalmology, 2017, 101, 1298-1302.	3.9	8
205	NEW OBSERVATIONS REGARDING THE RETINOPATHY OF GENETICALLY CONFIRMED KEARNSâ€“SAYRE SYNDROME. Retinal Cases and Brief Reports, 2018, 12, 349-358.	0.6	8
206	A novel, pathogenic dinucleotide deletion in the mitochondrial MT-TY gene causing myasthenia-like features. Neuromuscular Disorders, 2020, 30, 661-668.	0.6	8
207	Earlyâ€onset coenzyme Q10 deficiency associated with ataxia and respiratory chain dysfunction due to novel pathogenic COQ8A variants, including a large intragenic deletion. JIMD Reports, 2020, 54, 45-53.	1.5	8
208	PPA2-associated sudden cardiac death: extending the clinical and allelic spectrum in 20 new families. Genetics in Medicine, 2021, 23, 2415-2425.	2.4	8
209	The Effect of tRNA[Ser]Sec Isopentenylation on Selenoprotein Expression. International Journal of Molecular Sciences, 2021, 22, 11454.	4.1	8
210	Identification of a novel heterozygous guanosine monophosphate reductase (GMPR) variant in a patient with a lateâ€onset disorder of mitochondrial DNA maintenance. Clinical Genetics, 2020, 97, 276-286.	2.0	7
211	A novel de novo ACTA1 variant in a patient with nemaline myopathy and mitochondrial Complex I deficiency. Neuromuscular Disorders, 2020, 30, 159-164.	0.6	7
212	ATG7 safeguards human neural integrity. Autophagy, 2021, 17, 2651-2653.	9.1	7
213	Identification and characterization of novel <scp>MPC1</scp> gene variants causing mitochondrial pyruvate carrier deficiency. Journal of Inherited Metabolic Disease, 2022, 45, 264-277.	3.6	7
214	Periventricular Calcification, Abnormal Pterins and Dry Thickened Skin: Expanding the Clinical Spectrum of RMND1?. JIMD Reports, 2015, 26, 13-19.	1.5	6
215	Clinical features of the pathogenic m.5540G>A mitochondrial transfer RNA tryptophan gene mutation. Neuromuscular Disorders, 2016, 26, 702-705.	0.6	6
216	Machine learning algorithms reveal the secrets of mitochondrial dynamics. EMBO Molecular Medicine, 2021, 13, e14316.	6.9	6

#	Article	IF	Citations
217	RRM1 variants cause a mitochondrial DNA maintenance disorder via impaired de novo nucleotide synthesis. Journal of Clinical Investigation, 2022, 132, .	8.2	6
218	The swinging pendulum of biomarkers in mitochondrial disease. Neurology, 2016, 87, 2286-2287.	1.1	5
219	Sideroblastic anemia with myopathy secondary to novel, pathogenic missense variants in the YARS2 gene. Haematologica, 2018, 103, e564-e566.	3.5	5
220	A novel MT-CO2 variant causing cerebellar ataxia and neuropathy: The role of muscle biopsy in diagnosis and defining pathogenicity. Neuromuscular Disorders, 2021, 31, 1186-1193.	0.6	5
221	Characterising a homozygous twoâ€exon deletion in UQCRH : comparing human and mouse phenotypes. EMBO Molecular Medicine, 2021, 13, e14397.	6.9	5
222	The application of Raman spectroscopy to the diagnosis of mitochondrial muscle disease: A preliminary comparison between fibre optic probe and microscope formats. Journal of Raman Spectroscopy, 2022, 53, 172-181.	2.5	5
223	A novel pathogenic m.4412G>A MT-TM mitochondrial DNA variant associated with childhood-onset seizures, myopathy and bilateral basal ganglia changes. Mitochondrion, 2019, 47, 18-23.	3.4	4
224	The feasibility of muscle mitochondrial respiratory chain phenotyping across the cognitive spectrum in Parkinson's disease. Experimental Gerontology, 2020, 138, 110997.	2.8	4
225	Progressive external ophthalmoplegia due to a recurrent de novo m.15990C>T MT-TP (mt-tRNAPro) gene variant. Neuromuscular Disorders, 2020, 30, 346-350.	0.6	4
226	Clinical and Pathological Features of Mitochondrial DNA Deletion Disease Following Antiretroviral Treatment. JAMA Neurology, 2015, 72, 603.	9.0	3
227	Camptocormia and shuffling gait due to a novel MT-TV mutation: Diagnostic pitfalls. Neurology: Genetics, 2017, 3, e147.	1.9	3
228	Pathogenic SLC25A26 variants impair SAH transport activity causing mitochondrial disease. Human Molecular Genetics, 2022, 31, 2049-2062.	2.9	3
229	Biallelic variants in TAMM41 are associated with low muscle cardiolipin levels, leading to neonatal mitochondrial disease. Human Genetics and Genomics Advances, 2022, 3, 100097.	1.7	3
230	Novel POLG variants associated with late-onset de novo status epilepticus and progressive ataxia. Neurology: Genetics, 2017, 3, e181.	1.9	2
231	A novel mitochondrial m.4414T>C MT-TM gene variant causing progressive external ophthalmoplegia and myopathy. Neuromuscular Disorders, 2019, 29, 693-697.	0.6	2
232	Multisystem mitochondrial disease caused by a rare m.10038G>A mitochondrial tRNA ^{Gly} (MT-TG) variant. Neurology: Genetics, 2020, 6, e413.	1.9	2
233	Opening One's Eyes to Mosaicism in Progressive External Ophthalmoplegia. Neurology: Genetics, 2017, 3, e202.	1.9	1
234	The m.15043GÂ>ÂA MT-CYB variant is not a pathogenic mtDNA variant. Journal of the Neurological Sciences, 2020, 417, 116950.	0.6	1

#	Article	IF	Citations
235	Albinism and a mitochondrial DNA deletion. Ophthalmic Genetics, 2020, 41, 295-298.	1.2	1
236	Uniparental isodisomy of chromosome 2 causing MRPL44-related multisystem mitochondrial disease. Molecular Biology Reports, 2021, 48, 2093-2104.	2.3	1
237	Nonrandom tissue distribution of mutant mtDNA. , 1999, 85, 498.		1
238	Lateâ€onset optic atrophy, ataxia, and myopathy associated with a mutation of a complex II gene. Annals of Neurology, 2000, 48, 330-335.	5.3	1
239	P35â€…Histiocytoid cardiomyopathy and microphthalmia with linear skin defects syndrome: Phenotypes linked by truncating variants inNDUFB11. Heart, 2016, 102, A18.2-A18.	2.9	0
240	Cystic Leukoencephalopathy due to NDUFV1 mutationâ€”A Report of the Phenotype and Its Rare Co-occurrence with Primary Hyperoxaluria. Journal of Pediatric Neurology, 2016, 14, 126-132.	0.2	0
241	Nuclear genetic disorders of mitochondrial DNA gene expression. , 2020, , 375-409.		0