Robert Mcfarland, Frcpch

List of Publications by Year
in descending order

178 papers	11,555 citations	28190 55 h-index	33814 99 g-index
189 all docs	189 docs citations	189 times ranked	10365 citing authors

#	Article	IF	Citations
1	Forecasting stroke-like episodes and outcomes in mitochondrial disease. Brain, 2022, 145, 542-554.	3.7	25
2	Natural History of Leigh Syndrome: A Study of Disease Burden and Progression. Annals of Neurology, 2022, 91, 117-130.	2.8	17
3	Identification and characterization of novel <scp>MPC1</scp> gene variants causing mitochondrial pyruvate carrier deficiency. Journal of Inherited Metabolic Disease, 2022, 45, 264-277.	1.7	7
4	COVID-19â€"Related Outcomes in Primary Mitochondrial Diseases. Neurology, 2022, 98, 576-582.	1.5	7
5	Biallelic variants in TAMM41 are associated with low muscle cardiolipin levels, leading to neonatal mitochondrial disease. Human Genetics and Genomics Advances, 2022, 3, 100097.	1.0	3
6	Community Consensus Guidelines to Support FAIR Data Standards in Clinical Research Studies in Primary Mitochondrial Disease. Genetics & Genomics Next, 2022, 3, 2100047.	0.8	1
7	<scp>l</scp> -Arginine in Mitochondrial Encephalopathy, Lactic Acidosis, and Stroke-like Episodes. Neurology, 2022, 98, .	1.5	15
8	Phase II Feasibility Study of the Efficacy, Tolerability, and Impact on the Gut Microbiome of a Low-Residue (Fiber) Diet in Adult Patients With Mitochondrial Disease. , 2022, 1, 666-677.		0
9	Delineating selective vulnerability of inhibitory interneurons in Alpers' syndrome. Neuropathology and Applied Neurobiology, 2022, 48, .	1.8	8
10	Neuromuscular Junction Abnormalities in Mitochondrial Disease. Neurology: Clinical Practice, 2021, 11, 97-104.	0.8	10
11	Comment on â€œA severe linezolidâ€induced rhabdomyolysis and lactic acidosis in Leigh syndromeâ€. Journal of Inherited Metabolic Disease, 2021, 44, 6-7.	1.7	2
12	An international classification of inherited metabolic disorders (<scp>ICIMD</scp>). Journal of Inherited Metabolic Disease, 2021, 44, 164-177.	1.7	146
13	The molecular pathology of pathogenic mitochondrial tRNA variants. FEBS Letters, 2021, 595, 1003-1024.	1.3	29
14	Current and Emerging Clinical Treatment in Mitochondrial Disease. Molecular Diagnosis and Therapy, 2021, 25, 181-206.	1.6	36
15	Risk of cardiac manifestations in adult mitochondrial disease caused by nuclear genetic defects. Open Heart, 2021, 8, e001510.	0.9	3
16	Developmental Consequences of Defective ATG7-Mediated Autophagy in Humans. New England Journal of Medicine, 2021, 384, 2406-2417.	13.9	84
17	Mitochondrial disease in adults: recent advances and future promise. Lancet Neurology, The, 2021, 20, 573-584.	4.9	96
18	Toning up but powering down: does mitochondrial dysfunction lead to loss of ambulation in cerebral palsy?. Developmental Medicine and Child Neurology, 2021, 63, 1136-1136.	1.1	0

#	Article	IF	Citations
19	PPA2-associated sudden cardiac death: extending the clinical and allelic spectrum in 20 new families. Genetics in Medicine, 2021, 23, 2415-2425.	1.1	8
20	The Effect of tRNA[Ser]Sec Isopentenylation on Selenoprotein Expression. International Journal of Molecular Sciences, 2021, 22, 11454.	1.8	8
21	Recent advances in understanding the molecular genetic basis of mitochondrial disease. Journal of Inherited Metabolic Disease, 2020, 43, 36-50.	1.7	113
22	Congenital myasthenic syndrome with mild intellectual disability caused by a recurrent SLC25A1 variant. European Journal of Human Genetics, 2020, 28, 373-377.	1.4	20
23	Pathogenic Bi-allelic Mutations in NDUFAF8 Cause Leigh Syndrome with an Isolated Complex I Deficiency. American Journal of Human Genetics, 2020, 106, 92-101.	2.6	39
24	A novel de novo ACTA1 variant in a patient with nemaline myopathy and mitochondrial Complex I deficiency. Neuromuscular Disorders, 2020, 30, 159-164.	0.3	7
25	A novel, pathogenic dinucleotide deletion in the mitochondrial MT-TY gene causing myasthenia-like features. Neuromuscular Disorders, 2020, 30, 661-668.	0.3	8
26	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.	0.7	8
27	SURF1 related Leigh syndrome: Clinical and molecular findings of 16 patients from Turkey. Molecular Genetics and Metabolism Reports, 2020, 25, 100657.	0.4	10
28	Lower urinary tract dysfunction in adult patients with mitochondrial disease. Neurourology and Urodynamics, 2020, 39, 2253-2263.	0.8	5
29	Nuclear genetic disorders of mitochondrial DNA gene expression. , 2020, , 375-409.		0
30	The genetic basis of isolated mitochondrial complex II deficiency. Molecular Genetics and Metabolism, 2020, 131, 53-65.	0.5	22
31	Ultrasensitive deletion detection links mitochondrial DNA replication, disease, and aging. Genome Biology, 2020, 21, 248.	3.8	48
32	The m.15043GÂ>ÂA MT-CYB variant is not a pathogenic mtDNA variant. Journal of the Neurological Sciences, 2020, 417, 116950.	0.3	1
33	Safety of drug use in patients with a primary mitochondrial disease: An international Delphiâ€based consensus. Journal of Inherited Metabolic Disease, 2020, 43, 800-818.	1.7	42
34	Measuring the effects of exercise in neuromuscular disorders: a systematic review and meta-analyses. Wellcome Open Research, 2020, 5, 84.	0.9	19
35	Biâ€allelic pathogenic variants in NDUFC2 cause earlyâ€onset Leigh syndrome and stalled biogenesis of complex I. EMBO Molecular Medicine, 2020, 12, e12619.	3.3	17
36	Instability of the mitochondrial alanyl-tRNA synthetase underlies fatal infantile-onset cardiomyopathy. Human Molecular Genetics, 2019, 28, 258-268.	1.4	19

#	Article	IF	Citations
37	Dissecting the neuronal vulnerability underpinning Alpersâ€™ syndrome: a clinical and neuropathological study. Brain Pathology, 2019, 29, 97-113.	2.1	20
38	Resolving complexity in mitochondrial disease: Towards precision medicine. Molecular Genetics and Metabolism, 2019, 128, 19-29.	0.5	25
39	Diagnosis of â€˜possibleâ€™ mitochondrial disease: an existential crisis. Journal of Medical Genetics, 2019, 56, 123-130.	1.5	42
40	Cognitive deficits in adult m.3243A>Gâ€•and m.8344A>Gâ€•related mitochondrial disease: importance of correcting for baseline intellectual ability. Annals of Clinical and Translational Neurology, 2019, 6, 826-836.	1.7	10
41	Pathogenic variants in MTâ€ATP6: A United Kingdomâ€“based mitochondrial disease cohort study. Annals of Neurology, 2019, 86, 310-315.	2.8	33
42	Surveillance for variant CJD: should more children with neurodegenerative diseases have autopsies?. Archives of Disease in Childhood, 2019, 104, 360-365.	1.0	7
43	Mitochondrial Donation â€” Which Women Could Benefit?. New England Journal of Medicine, 2019, 380, 1971-1972.	13.9	25
44	Mutations inELAC2associated with hypertrophic cardiomyopathy impair mitochondrial tRNA 3â€²â€•end processing. Human Mutation, 2019, 40, 1731-1748.	1.1	31
45	Leigh Syndrome. , 2019, , 151-167.		0
46	Height as a Clinical Biomarker of Disease Burden in Adult Mitochondrial Disease. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 2057-2066.	1.8	19
47	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.	1.6	4
48	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.	1.7	17
49	Copy-choice recombination during mitochondrial L-strand synthesis causes DNA deletions. Nature Communications, 2019, 10, 759.	5.8	34
50	Consensus-based statements for the management of mitochondrial stroke-like episodes. Wellcome Open Research, 2019, 4, 201.	0.9	66
51	MT-ND5 Mutation Exhibits Highly Variable Neurological Manifestations at Low Mutant Load. EBioMedicine, 2018, 30, 86-93.	2.7	47
52	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. American Journal of Human Genetics, 2018, 102, 494-504.	2.6	59
53	The adjunctive application of transcranial direct current stimulation in the management of de novo refractory epilepsia partialis continua in adolescentâ€onset <scp>POLG</scp>â€related mitochondrial disease. Epilepsia Open, 2018, 3, 103-108.	1.3	16
54	Overview of Approaches to Mitochondrial Disease Therapy. FIRE Forum for International Research in Education, 2018, 6, 232640981775296.	0.7	6

#	Article	IF	Citations
55	Pathological mechanisms underlying single largeâ€scale mitochondrial <scp>DNA</scp> deletions. Annals of Neurology, 2018, 83, 115-130.	2.8	42
56	Clinical, biochemical, and genetic features associated with VARS2 -related mitochondrial disease. Human Mutation, 2018, 39, 563-578.	1.1	22
57	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.	1.1	21
58	Phenotypic heterogeneity in m.3243A>G mitochondrial disease: The role of nuclear factors. Annals of Clinical and Translational Neurology, 2018, 5, 333-345.	1.7	102
59	Scientific and Ethical Issues in Mitochondrial Donation. New Bioethics, 2018, 24, 57-73.	0.5	25
60	Retrospective natural history of thymidine kinase 2 deficiency. Journal of Medical Genetics, 2018, 55, 515-521.	1.5	73
61	The genotypic and phenotypic spectrum of MTO1 deficiency. Molecular Genetics and Metabolism, 2018, 123, 28-42.	0.5	24
62	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.	2.6	41
63	Mitochondrial donation: from test tube to clinic. Lancet, The, 2018, 392, 1191-1192.	6.3	30
64	Mutations of the mitochondrial carrier translocase channel subunit TIM22 cause early-onset mitochondrial myopathy. Human Molecular Genetics, 2018, 27, 4135-4144.	1.4	30
65	<scp>OXA</scp> 1L mutations cause mitochondrial encephalopathy and a combined oxidative phosphorylation defect. EMBO Molecular Medicine, 2018, 10, .	3.3	54
66	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. Orphanet Journal of Rare Diseases, 2018, 13, 120.	1.2	61
67	Outcome measures for children with mitochondrial disease: consensus recommendations for future studies from a Delphiâ€based international workshop. Journal of Inherited Metabolic Disease, 2018, 41, 1267-1273.	1.7	24
68	mt <scp>DNA</scp> heteroplasmy level and copy number indicate disease burden in m.3243A>G mitochondrial disease. EMBO Molecular Medicine, 2018, 10, .	3.3	199
69	Clinical Features, Molecular Heterogeneity, and Prognostic Implications in YARS2-Related Mitochondrial Myopathy. JAMA Neurology, 2017, 74, 686.	4.5	41
70	De novo mtDNA point mutations are common and have a low recurrence risk. Journal of Medical Genetics, 2017, 54, 73-83.	1.5	54
71	De novo CTBP1 variant is associated with decreased mitochondrial respiratory chain activities. Neurology: Genetics, 2017, 3, e187.	0.9	11
72	Novel GFM2 variants associated with early-onset neurological presentations of mitochondrial disease and impaired expression of OXPHOS subunits. Neurogenetics, 2017, 18, 227-235.	0.7	10

#	Article	IF	Citations
73	The presence of anaemia negatively influences survival in patients with POLG disease. Journal of Inherited Metabolic Disease, 2017, 40, 861-866.	1.7	8
74	Decreased male reproductive success in association with mitochondrial dysfunction. European Journal of Human Genetics, 2017, 25, 1162-1164.	1.4	18
75	International Workshop:. Neuromuscular Disorders, 2017, 27, 1126-1137.	0.3	58
76	Using a quantitative quadruple immunofluorescent assay to diagnose isolated mitochondrial Complex I deficiency. Scientific Reports, 2017, 7, 15676.	1.6	20
77	RMND1-Related Leukoencephalopathy With Temporal Lobe Cysts and Hearing Lossâ€"Another Mendelian Mimicker of Congenital Cytomegalovirus Infection. Pediatric Neurology, 2017, 66, 59-62.	1.0	12
78	Pathogenic variants in HTRA2 cause an earlyâ€onset mitochondrial syndrome associated with 3â€methylglutaconic aciduria. Journal of Inherited Metabolic Disease, 2017, 40, 121-130.	1.7	23
79	Compound heterozygous RMND1 gene variants associated with chronic kidney disease, dilated cardiomyopathy and neurological involvement: a case report. BMC Research Notes, 2016, 9, 325.	0.6	15
80	The clinical, biochemical and genetic features associated withRMND1-related mitochondrial disease. Journal of Medical Genetics, 2016, 53, 768-775.	1.5	35
81	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.	0.9	40
82	Clinical, Genetic, and Radiological Features of Extrapyramidal Movement Disorders in Mitochondrial Disease. JAMA Neurology, 2016, 73, 668.	4.5	69
83	Nutritional interventions in primary mitochondrial disorders: Developing an evidence base. Molecular Genetics and Metabolism, 2016, 119, 187-206.	0.5	41
84	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.	2.6	93
85	Teaching Neuro Images : Neuroradiologic evolution of Leigh disease. Neurology, 2016, 87, e159-e160.	1.5	0
86	Epilepsy due to mutations in the mitochondrial polymerase gamma (<scp>POLG</scp>) gene: A clinical and molecular genetic review. Epilepsia, 2016, 57, 1531-1545.	2.6	58
87	Sudden Cardiac Death Due to Deficiency of the Mitochondrial Inorganic Pyrophosphatase PPA2. American Journal of Human Genetics, 2016, 99, 674-682.	2.6	48
88	The Spectrum of Mitochondrial Ultrastructural Defects in Mitochondrial Myopathy. Scientific Reports, 2016, 6, 30610.	1.6	165
89	International Paediatric Mitochondrial Disease Scale. Journal of Inherited Metabolic Disease, 2016, 39, 705-712.	1.7	16
90	Pseudoâ€obstruction, stroke, and mitochondrial dysfunction: A lethal combination. Annals of Neurology, 2016, 80, 686-692.	2.8	40

#	Article	IF	Citations
91	Mitochondrial diseases. Nature Reviews Disease Primers, 2016, 2, 16080.	18.1	1,001
92	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.	1.5	31
93	Recessive Mutations in TRMT10C Cause Defects in Mitochondrial RNA Processing and Multiple Respiratory Chain Deficiencies. American Journal of Human Genetics, 2016, 98, 993-1000.	2.6	89
94	Solid organ transplantation in primary mitochondrial disease: Proceed with caution. Molecular Genetics and Metabolism, 2016, 118, 178-184.	0.5	55
95	Both mitochondrial DNA and mitonuclear gene mutations cause hearing loss through cochlear dysfunction. Brain, 2016, 139, e33-e33.	3.7	15
96	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.	1.0	53
97	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.	1.7	79
98	Epilepsy in adults with mitochondrial disease: A cohort study. Annals of Neurology, 2015, 78, 949-957.	2.8	62
99	Preliminary Evaluation of Clinician Rated Outcome Measures in Mitochondrial Disease. Journal of Neuromuscular Diseases, 2015, 2, 151-155.	1.1	8
100	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
101	Prevalence of nuclear and mitochondrial <scp>DNA</scp> mutations related to adult mitochondrial disease. Annals of Neurology, 2015, 77, 753-759.	2.8	706
102	Clonal Expansion of Secondary Mitochondrial DNA Deletions Associated With Spinocerebellar Ataxia Type 28. JAMA Neurology, 2015, 72, 106.	4.5	41
103	LRPPRCmutations cause early-onset multisystem mitochondrial disease outside of the French-Canadian population. Brain, 2015, 138, 3503-3519.	3.7	81
104	Causes of Death in Adults with Mitochondrial Disease. JIMD Reports, 2015, 26, 103-113.	0.7	36
105	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.	0.9	31
106	Mitochondrial Donation â€" How Many Women Could Benefit?. New England Journal of Medicine, 2015, 372, 885-887.	13.9	87
107	Novel MTND1 mutations cause isolated exercise intolerance, complex I deficiency and increased assembly factor expression. Clinical Science, 2015, 128, 895-904.	1.8	21
108	Perceived fatigue is highly prevalent and debilitating in patients with mitochondrial disease. Neuromuscular Disorders, 2015, 25, 563-566.	0.3	67

#	Article	IF	Citations
109	Long-term survival in a child with severe encephalopathy, multiple respiratory chain deficiency and GFM1 mutations. Frontiers in Genetics, 2015, 6, 102.	1.1	13
110	The GABA Transaminase, ABAT, Is Essential for Mitochondrial Nucleoside Metabolism. Cell Metabolism, 2015, 21, 417-427.	7.2	119
111	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.	1.7	45
112	A CLINICAL AUDIT OF ACUTE MANAGEMENT OF STROKE-LIKE EPISODES FROM A NATIONAL MITOCHONDRIAL DISEASE CENTRE. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, e4.161-e4.	0.9	0
113	A truncating PET100 variant causing fatal infantile lactic acidosis and isolated cytochrome c oxidase deficiency. European Journal of Human Genetics, 2015, 23, 935-939.	1.4	32
114	The urinary proteome and metabonome differ from normal in adults with mitochondrial disease. Kidney International, 2015, 87, 610-622.	2.6	41
115	A national perspective on prenatal testing for mitochondrial disease. European Journal of Human Genetics, 2014, 22, 1255-1259.	1.4	57
116	Defective i6A37 Modification of Mitochondrial and Cytosolic tRNAs Results from Pathogenic Mutations in TRIT1 and Its Substrate tRNA. PLoS Genetics, 2014, 10, e1004424.	1.5	112
117	Mitochondrial myopathies in adults and children. Current Opinion in Neurology, 2014, 27, 576-582.	1.8	32
118	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.	3.8	304
119	Discrete gait characteristics are associated with m.3243A>G and m.8344A>G variants of mitochondrial disease and its pathological consequences. Journal of Neurology, 2014, 261, 73-82.	1.8	11
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.	1.8	61
121	Disease progression in patients with single, large-scale mitochondrial DNA deletions. Brain, 2014, 137, 323-334.	3.7	103
122	Mutations in the SPG7 gene cause chronic progressive external ophthalmoplegia through disordered mitochondrial DNA maintenance. Brain, 2014, 137, 1323-1336.	3.7	151
123	SURF1 deficiency: a multi-centre natural history study. Orphanet Journal of Rare Diseases, 2013, 8, 96.	1.2	107
124	The m.3291T>C mt-tRNALeu(UUR) mutation is definitely pathogenic and causes multisystem mitochondrial disease. Journal of the Neurological Sciences, 2013, 325, 165-169.	0.3	7
125	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.	1.1	62
126	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.	0.9	193

#	Article	IF	Citations
127	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
128	Mutations in FBXL4 Cause Mitochondrial Encephalopathy and a Disorder of Mitochondrial DNA Maintenance. American Journal of Human Genetics, 2013, 93, 471-481.	2.6	137
129	New treatments for mitochondrial diseaseâ€"no time to drop our standards. Nature Reviews Neurology, 2013, 9, 474-481.	4.9	157
130	Cerebral folate deficiency--mishaps and misdirection. Brain, 2012, 135, 2002-2003.	3.7	9
131	Adults with RRM2B-related mitochondrial disease have distinct clinical and molecular characteristics. Brain, 2012, 135, 3392-3403.	3.7	70
132	Mutations in the mitochondrial tRNASer(AGY) gene are associated with deafness, retinal degeneration, myopathy and epilepsy. European Journal of Human Genetics, 2012, 20, 897-904.	1.4	10
133	Recessive germlineSDHAandSDHBmutations causing leukodystrophy and isolated mitochondrial complex II deficiency. Journal of Medical Genetics, 2012, 49, 569-577.	1.5	100
134	Cerebellar Ataxia in Patients With Mitochondrial DNA Disease. Journal of Neuropathology and Experimental Neurology, 2012, 71, 148-161.	0.9	91
135	Cardiomyopathy is common in patients with the mitochondrial DNA m.3243A>G mutation and correlates with mutation load. Neuromuscular Disorders, 2012, 22, 592-596.	0.3	34
136	MPV17 mutation causes neuropathy and leukoencephalopathy with multiple mtDNA deletions in muscle. Neuromuscular Disorders, 2012, 22, 587-591.	0.3	51
137	The clinical spectrum of the m.10191T>C mutation in complex Iâ€deficient Leigh syndrome. Developmental Medicine and Child Neurology, 2012, 54, 500-506.	1.1	35
138	Mitochondrial respiratory chain disease in children undergoing cardiac transplantation: A prospective study. International Journal of Cardiology, 2012, 155, 305-306.	0.8	14
139	A proposed consensus panel of organisms for determining evolutionary conservation of mt-tRNA point mutations. Mitochondrion, 2012, 12, 533-538.	1.6	28
140	Toward a mtDNA locus-specific mutation database using the LOVD platform. Human Mutation, 2012, 33, 1352-1358.	1.1	8
141	Prevalence and severity of voice and swallowing difficulties in mitochondrial disease. International Journal of Language and Communication Disorders, 2012, 47, 106-111.	0.7	8
142	Rapid-onset, linezolid-induced lactic acidosis in MELAS. Mitochondrion, 2011, 11, 992-993.	1.6	25
143	Respiratory chain complex I deficiency caused by mitochondrial DNA mutations. European Journal of Human Genetics, 2011, 19, 769-775.	1.4	102
144	Autosomal dominant acute necrotising encephalopathy: AÂcase report with possible disease-expression modification by coincidental homocysteinuria. European Journal of Paediatric Neurology, 2011, 15, 174-176.	0.7	4

#	Article	IF	Citations
145	Nijmegen paediatric CDG rating scale: a novel tool to assess disease progression. Journal of Inherited Metabolic Disease, 2011, 34, 923-927.	1.7	50
146	A comparative analysis approach to determining the pathogenicity of mitochondrial tRNA mutations. Human Mutation, 2011, 32, 1319-1325.	1.1	159
147	Nuclear factors involved in mitochondrial translation cause a subgroup of combined respiratory chain deficiency. Brain, 2011, 134, 183-195.	3.7	66
148	Maternally inherited mitochondrial DNA disease in consanguineous families. European Journal of Human Genetics, 2011, 19, 1226-1229.	1.4	20
149	mtDNA disease for the neurologist. Future Neurology, 2011, 6, 63-80.	0.9	0
150	The clinical presentation of mitochondrial diseases in children with progressive intellectual and neurological deterioration: a national, prospective, populationâ€based study. Developmental Medicine and Child Neurology, 2010, 52, 434-440.	1.1	35
151	Mitochondrial tRNA mutations and disease. Wiley Interdisciplinary Reviews RNA, 2010, 1, 304-324.	3.2	145
152	A neurological perspective on mitochondrial disease. Lancet Neurology, The, 2010, 9, 829-840.	4.9	316
153	Mitochondrial diseases in childhood: a clinical approach to investigation and management. Developmental Medicine and Child Neurology, 2010, 52, 422-433.	1.1	56
154	Unilateral Horner's syndrome: An unusual childhood presentation. Journal of Paediatrics and Child Health, 2010, 46, 695-695.	0.4	1
155	The p.M292T NDUFS2 mutation causes complex I-deficient Leigh syndrome in multiple families. Brain, 2010, 133, 2952-2963.	3.7	69
156	The pathogenic m.3243A>T mitochondrial DNA mutation is associated with a variable neurological phenotype. Neuromuscular Disorders, 2010, 20, 403-406.	0.3	7
157	Long-term survival of neonatal mitochondrial complex III deficiency associated with a novel BCS1L gene mutation. Molecular Genetics and Metabolism, 2010, 100, 345-348.	0.5	27
158	Molecular basis of infantile reversible cytochrome c oxidase deficiency myopathy. Brain, 2009, 132, 3165-3174.	3.7	112
159	Pathogenic mitochondrial tRNA mutations - Which mutations are inherited and why?. Human Mutation, 2009, 30, E984-E992.	1.1	49
160	Neuromuscular disease presentation with three genetic defects involving two genomes. Neuromuscular Disorders, 2009, 19, 841-844.	0.3	6
161	Prevalence of mitochondrial DNA disease in adults. Annals of Neurology, 2008, 63, 35-39.	2.8	540
162	The m.5650G>A mitochondrial tRNAAla mutation is pathogenic and causes a phenotype of pure myopathy. Neuromuscular Disorders, 2008, 18, 63-67.	0.3	32

#	Article	IF	Citations
163	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.	3.7	381
164	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.	6.5	74
165	Diabetes and Deafness: Is it sufficient to screen for the mitochondrial 3243A>G mutation alone?. Diabetes Care, 2007, 30, 2238-2239.	4.3	10
166	Sporadic myopathy and exercise intolerance associated with the mitochondrial 8328G>A tRNALys mutation. Journal of Neurology, 2007, 254, 1283-1285.	1.8	15
167	Phenotypic spectrum associated with mutations of the mitochondrial polymerase Â gene. Brain, 2006, 129, 1674-1684.	3.7	397
168	Noninvasive diagnosis of the 3243A>G mitochondrial DNA mutation using urinary epithelial cells. European Journal of Human Genetics, 2004, 12, 778-781.	1.4	107
169	Assigning pathogenicity to mitochondrial tRNA mutations: when â€˜definitely maybeâ€™ is not good enough. Trends in Genetics, 2004, 20, 591-596.	2.9	159
170	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.	2.8	164
171	Familial myopathy: New insights into the T14709C mitochondrial tRNA mutation. Annals of Neurology, 2004, 55, 478-484.	2.8	71
172	Childhood neurological presentation of a novel mitochondrial tRNAVal gene mutation. Journal of the Neurological Sciences, 2004, 225, 99-103.	0.3	18
173	A novel sporadic mutation in cytochrome c oxidase subunit II as a cause of rhabdomyolysis. Neuromuscular Disorders, 2004, 14, 162-166.	0.3	58
174	The diagnosis of mitochondrial muscle disease. Neuromuscular Disorders, 2004, 14, 237-245.	0.3	178
175	A novel mitochondrial DNA tRNAIle (A4267G) mutation in a sporadic patient with mitochondrial myopathy. Neuromuscular Disorders, 2002, 12, 659-664.	0.3	20
176	The neurology of mitochondrial DNA disease. Lancet Neurology, The, 2002, 1, 343-351.	4.9	115
177	Multiple neonatal deaths due to a homoplasmic mitochondrial DNA mutation. Nature Genetics, 2002, 30, 145-146.	9.4	162
178	Mitochondrial disorders. , 0, , 188-211.		0