

# Robert Mcfarland, Frcp

## List of Publications by Year in descending order

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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	Mitochondrial diseases. Nature Reviews Disease Primers, 2016, 2, 16080.	18.1	1,001
2	Prevalence of nuclear and mitochondrial <scp>DNA</scp> mutations related to adult mitochondrial disease. Annals of Neurology, 2015, 77, 753-759.	2.8	706
3	Prevalence of mitochondrial DNA disease in adults. Annals of Neurology, 2008, 63, 35-39.	2.8	540
4	Phenotypic spectrum associated with mutations of the mitochondrial polymerase $\gamma$ gene. Brain, 2006, 129, 1674-1684.	3.7	397
5	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
6	A neurological perspective on mitochondrial disease. Lancet Neurology, The, 2010, 9, 829-840.	4.9	316
7	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
8	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
9	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
10	The diagnosis of mitochondrial muscle disease. Neuromuscular Disorders, 2004, 14, 237-245.	0.3	178
11	The Spectrum of Mitochondrial Ultrastructural Defects in Mitochondrial Myopathy. Scientific Reports, 2016, 6, 30610.	1.6	165
12	De novo mutations in the mitochondrial ND3 gene as a cause of infantile mitochondrial encephalopathy and complex I deficiency. Annals of Neurology, 2004, 55, 58-64.	2.8	164
13	Multiple neonatal deaths due to a homoplasmic mitochondrial DNA mutation. Nature Genetics, 2002, 30, 145-146.	9.4	162
14	Assigning pathogenicity to mitochondrial tRNA mutations: when “definitely maybe”™ is not good enough. Trends in Genetics, 2004, 20, 591-596.	2.9	159
15	A comparative analysis approach to determining the pathogenicity of mitochondrial tRNA mutations. Human Mutation, 2011, 32, 1319-1325.	1.1	159
16	New treatments for mitochondrial disease—no time to drop our standards. Nature Reviews Neurology, 2013, 9, 474-481.	4.9	157
17	Mutations in the SPG7 gene cause chronic progressive external ophthalmoplegia through disordered mitochondrial DNA maintenance. Brain, 2014, 137, 1323-1336.	3.7	151
18	An international classification of inherited metabolic disorders (<scp>ICIMD</scp>). Journal of Inherited Metabolic Disease, 2021, 44, 164-177.	1.7	146

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19	Mitochondrial tRNA mutations and disease. <i>Wiley Interdisciplinary Reviews RNA</i> , 2010, 1, 304-324.	3.2	145
20	Mutations in FBXL4 Cause Mitochondrial Encephalopathy and a Disorder of Mitochondrial DNA Maintenance. <i>American Journal of Human Genetics</i> , 2013, 93, 471-481.	2.6	137
21	The GABA Transaminase, ABAT, Is Essential for Mitochondrial Nucleoside Metabolism. <i>Cell Metabolism</i> , 2015, 21, 417-427.	7.2	119
22	The neurology of mitochondrial DNA disease. <i>Lancet Neurology</i> , The, 2002, 1, 343-351.	4.9	115
23	Recent advances in understanding the molecular genetic basis of mitochondrial disease. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 36-50.	1.7	113
24	Molecular basis of infantile reversible cytochrome c oxidase deficiency myopathy. <i>Brain</i> , 2009, 132, 3165-3174.	3.7	112
25	Defective i6A37 Modification of Mitochondrial and Cytosolic tRNAs Results from Pathogenic Mutations in TRIT1 and Its Substrate tRNA. <i>PLoS Genetics</i> , 2014, 10, e1004424.	1.5	112
26	Noninvasive diagnosis of the 3243A>G mitochondrial DNA mutation using urinary epithelial cells. <i>European Journal of Human Genetics</i> , 2004, 12, 778-781.	1.4	107
27	SURF1 deficiency: a multi-centre natural history study. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 96.	1.2	107
28	Disease progression in patients with single, large-scale mitochondrial DNA deletions. <i>Brain</i> , 2014, 137, 323-334.	3.7	103
29	Respiratory chain complex I deficiency caused by mitochondrial DNA mutations. <i>European Journal of Human Genetics</i> , 2011, 19, 769-775.	1.4	102
30	Phenotypic heterogeneity in m.3243A>G mitochondrial disease: The role of nuclear factors. <i>Annals of Clinical and Translational Neurology</i> , 2018, 5, 333-345.	1.7	102
31	Recessive germline <i>SDHA</i> and <i>SDHB</i> mutations causing leukodystrophy and isolated mitochondrial complex II deficiency. <i>Journal of Medical Genetics</i> , 2012, 49, 569-577.	1.5	100
32	Mitochondrial disease in adults: recent advances and future promise. <i>Lancet Neurology</i> , The, 2021, 20, 573-584.	4.9	96
33	Recurrent De Novo Dominant Mutations in SLC25A4 Cause Severe Early-Onset Mitochondrial Disease and Loss of Mitochondrial DNA Copy Number. <i>American Journal of Human Genetics</i> , 2016, 99, 860-876.	2.6	93
34	Cerebellar Ataxia in Patients With Mitochondrial DNA Disease. <i>Journal of Neuropathology and Experimental Neurology</i> , 2012, 71, 148-161.	0.9	91
35	Recessive Mutations in TRMT10C Cause Defects in Mitochondrial RNA Processing and Multiple Respiratory Chain Deficiencies. <i>American Journal of Human Genetics</i> , 2016, 98, 993-1000.	2.6	89
36	Mitochondrial Donation – How Many Women Could Benefit?. <i>New England Journal of Medicine</i> , 2015, 372, 885-887.	13.9	87

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37	Developmental Consequences of Defective ATG7-Mediated Autophagy in Humans. <i>New England Journal of Medicine</i> , 2021, 384, 2406-2417.	13.9	84
38	<i>LRPPRC</i> mutations cause early-onset multisystem mitochondrial disease outside of the French-Canadian population. <i>Brain</i> , 2015, 138, 3503-3519.	3.7	81
39	Succinateâ€CoA ligase deficiency due to mutations in <i>SUCLA2</i> and <i>SUCLG1</i>: phenotype and genotype correlations in 71 patients. <i>Journal of Inherited Metabolic Disease</i> , 2016, 39, 243-252.	1.7	79
40	Overexpression of human mitochondrial valyl tRNA synthetase can partially restore levels of cognate mt-tRNA <sup>Val</sup> carrying the pathogenic C25U mutation. <i>Nucleic Acids Research</i> , 2008, 36, 3065-3074.	6.5	74
41	Retrospective natural history of thymidine kinase 2 deficiency. <i>Journal of Medical Genetics</i> , 2018, 55, 515-521.	1.5	73
42	Familial myopathy: New insights into the T14709C mitochondrial tRNA mutation. <i>Annals of Neurology</i> , 2004, 55, 478-484.	2.8	71
43	Adults with RRM2B-related mitochondrial disease have distinct clinical and molecular characteristics. <i>Brain</i> , 2012, 135, 3392-3403.	3.7	70
44	The p.M292T NDUFS2 mutation causes complex I-deficient Leigh syndrome in multiple families. <i>Brain</i> , 2010, 133, 2952-2963.	3.7	69
45	Clinical, Genetic, and Radiological Features of Extrapyrimalidal Movement Disorders in Mitochondrial Disease. <i>JAMA Neurology</i> , 2016, 73, 668.	4.5	69
46	Perceived fatigue is highly prevalent and debilitating in patients with mitochondrial disease. <i>Neuromuscular Disorders</i> , 2015, 25, 563-566.	0.3	67
47	Nuclear factors involved in mitochondrial translation cause a subgroup of combined respiratory chain deficiency. <i>Brain</i> , 2011, 134, 183-195.	3.7	66
48	Consensus-based statements for the management of mitochondrial stroke-like episodes. <i>Wellcome Open Research</i> , 2019, 4, 201.	0.9	66
49	Pathogenic Mitochondrial t <sc>RNA</sc> Point Mutations: Nine Novel Mutations Affirm Their Importance as a Cause of Mitochondrial Disease. <i>Human Mutation</i> , 2013, 34, 1260-1268.	1.1	62
50	Epilepsy in adults with mitochondrial disease: A cohort study. <i>Annals of Neurology</i> , 2015, 78, 949-957.	2.8	62
51	Mutation of the human mitochondrial phenylalanine-tRNA synthetase causes infantile-onset epilepsy and cytochrome c oxidase deficiency. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2014, 1842, 56-64.	1.8	61
52	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 120.	1.2	61
53	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. <i>American Journal of Human Genetics</i> , 2018, 102, 494-504.	2.6	59
54	A novel sporadic mutation in cytochrome c oxidase subunit II as a cause of rhabdomyolysis. <i>Neuromuscular Disorders</i> , 2004, 14, 162-166.	0.3	58

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55	Epilepsy due to mutations in the mitochondrial polymerase gamma (<sc>POLG</sc>) gene: A clinical and molecular genetic review. <i>Epilepsia</i> , 2016, 57, 1531-1545.	2.6	58
56	International Workshop: Neuromuscular Disorders, 2017, 27, 1126-1137.	0.3	58
57	A national perspective on prenatal testing for mitochondrial disease. <i>European Journal of Human Genetics</i> , 2014, 22, 1255-1259.	1.4	57
58	Mitochondrial diseases in childhood: a clinical approach to investigation and management. <i>Developmental Medicine and Child Neurology</i> , 2010, 52, 422-433.	1.1	56
59	Solid organ transplantation in primary mitochondrial disease: Proceed with caution. <i>Molecular Genetics and Metabolism</i> , 2016, 118, 178-184.	0.5	55
60	De novo mtDNA point mutations are common and have a low recurrence risk. <i>Journal of Medical Genetics</i> , 2017, 54, 73-83.	1.5	54
61	<sc>OXA</sc> 1L mutations cause mitochondrial encephalopathy and a combined oxidative phosphorylation defect. <i>EMBO Molecular Medicine</i> , 2018, 10, .	3.3	54
62	Sudden adult death syndrome in m.3243A>G-related mitochondrial disease: an unrecognized clinical entity in young, asymptomatic adults. <i>European Heart Journal</i> , 2016, 37, 2552-2559.	1.0	53
63	MPV17 mutation causes neuropathy and leukoencephalopathy with multiple mtDNA deletions in muscle. <i>Neuromuscular Disorders</i> , 2012, 22, 587-591.	0.3	51
64	Nijmegen paediatric CDG rating scale: a novel tool to assess disease progression. <i>Journal of Inherited Metabolic Disease</i> , 2011, 34, 923-927.	1.7	50
65	Pathogenic mitochondrial tRNA mutations - Which mutations are inherited and why?. <i>Human Mutation</i> , 2009, 30, E984-E992.	1.1	49
66	A recessive homozygous p.Asp92Gly SDHD mutation causes prenatal cardiomyopathy and a severe mitochondrial complex II deficiency. <i>Human Genetics</i> , 2015, 134, 869-879.	1.8	49
67	Sudden Cardiac Death Due to Deficiency of the Mitochondrial Inorganic Pyrophosphatase PPA2. <i>American Journal of Human Genetics</i> , 2016, 99, 674-682.	2.6	48
68	Ultrasensitive deletion detection links mitochondrial DNA replication, disease, and aging. <i>Genome Biology</i> , 2020, 21, 248.	3.8	48
69	MT-ND5 Mutation Exhibits Highly Variable Neurological Manifestations at Low Mutant Load. <i>EBioMedicine</i> , 2018, 30, 86-93.	2.7	47
70	Clinical, morphological, biochemical, imaging and outcome parameters in 21 individuals with mitochondrial maintenance defect related to <sc>FBXL4</sc> mutations. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 905-914.	1.7	45
71	Defining cardiac adaptations and safety of endurance training in patients with m.3243A>G-related mitochondrial disease. <i>International Journal of Cardiology</i> , 2013, 168, 3599-3608.	0.8	43
72	Pathological mechanisms underlying single large-scale mitochondrial <sc>DNA</sc> deletions. <i>Annals of Neurology</i> , 2018, 83, 115-130.	2.8	42

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73	Diagnosis of “possible” mitochondrial disease: an existential crisis. <i>Journal of Medical Genetics</i> , 2019, 56, 123-130.	1.5	42
74	Safety of drug use in patients with a primary mitochondrial disease: An international Delphi-based consensus. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 800-818.	1.7	42
75	Clonal Expansion of Secondary Mitochondrial DNA Deletions Associated With Spinocerebellar Ataxia Type 28. <i>JAMA Neurology</i> , 2015, 72, 106.	4.5	41
76	The urinary proteome and metabonome differ from normal in adults with mitochondrial disease. <i>Kidney International</i> , 2015, 87, 610-622.	2.6	41
77	Nutritional interventions in primary mitochondrial disorders: Developing an evidence base. <i>Molecular Genetics and Metabolism</i> , 2016, 119, 187-206.	0.5	41
78	Clinical Features, Molecular Heterogeneity, and Prognostic Implications in <i>YARS2</i> -Related Mitochondrial Myopathy. <i>JAMA Neurology</i> , 2017, 74, 686.	4.5	41
79	Bi-allelic Mutations in <i>NDUFA6</i> Establish Its Role in Early-Onset Isolated Mitochondrial Complex I Deficiency. <i>American Journal of Human Genetics</i> , 2018, 103, 592-601.	2.6	41
80	Incidence of Primary Mitochondrial Disease in Children Younger Than 2 Years Presenting With Acute Liver Failure. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2016, 63, 592-597.	0.9	40
81	Pseudo-obstruction, stroke, and mitochondrial dysfunction: A lethal combination. <i>Annals of Neurology</i> , 2016, 80, 686-692.	2.8	40
82	Pathogenic Bi-allelic Mutations in <i>NDUFAF8</i> Cause Leigh Syndrome with an Isolated Complex I Deficiency. <i>American Journal of Human Genetics</i> , 2020, 106, 92-101.	2.6	39
83	Causes of Death in Adults with Mitochondrial Disease. <i>JIMD Reports</i> , 2015, 26, 103-113.	0.7	36
84	Current and Emerging Clinical Treatment in Mitochondrial Disease. <i>Molecular Diagnosis and Therapy</i> , 2021, 25, 181-206.	1.6	36
85	The clinical presentation of mitochondrial diseases in children with progressive intellectual and neurological deterioration: a national, prospective, population-based study. <i>Developmental Medicine and Child Neurology</i> , 2010, 52, 434-440.	1.1	35
86	The clinical spectrum of the m.10191T>C mutation in complex I-deficient Leigh syndrome. <i>Developmental Medicine and Child Neurology</i> , 2012, 54, 500-506.	1.1	35
87	The clinical, biochemical and genetic features associated with <i>RMND1</i> -related mitochondrial disease. <i>Journal of Medical Genetics</i> , 2016, 53, 768-775.	1.5	35
88	Cardiomyopathy is common in patients with the mitochondrial DNA m.3243A>G mutation and correlates with mutation load. <i>Neuromuscular Disorders</i> , 2012, 22, 592-596.	0.3	34
89	Copy-choice recombination during mitochondrial L-strand synthesis causes DNA deletions. <i>Nature Communications</i> , 2019, 10, 759.	5.8	34
90	Pathogenic variants in <i>MT-ATP6</i> : A United Kingdom-based mitochondrial disease cohort study. <i>Annals of Neurology</i> , 2019, 86, 310-315.	2.8	33

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91	The m.5650G>A mitochondrial tRNA <sup>Ala</sup> mutation is pathogenic and causes a phenotype of pure myopathy. <i>Neuromuscular Disorders</i> , 2008, 18, 63-67.	0.3	32
92	Mitochondrial myopathies in adults and children. <i>Current Opinion in Neurology</i> , 2014, 27, 576-582.	1.8	32
93	A truncating PET100 variant causing fatal infantile lactic acidosis and isolated cytochrome c oxidase deficiency. <i>European Journal of Human Genetics</i> , 2015, 23, 935-939.	1.4	32
94	Neuropathologic Characterization of Pontocerebellar Hypoplasia Type 6 Associated With Cardiomyopathy and Hydrops Fetalis and Severe Multisystem Respiratory Chain Deficiency due to Novel RARS2 Mutations. <i>Journal of Neuropathology and Experimental Neurology</i> , 2015, 74, 688-703.	0.9	31
95	A recurrent mitochondrial p.Trp22ArgNDUFB3 variant causes a distinctive facial appearance, short stature and a mild biochemical and clinical phenotype. <i>Journal of Medical Genetics</i> , 2016, 53, 634-641.	1.5	31
96	Mutations in ELAC2 associated with hypertrophic cardiomyopathy impair mitochondrial tRNA processing. <i>Human Mutation</i> , 2019, 40, 1731-1748.	1.1	31
97	Mitochondrial donation: from test tube to clinic. <i>Lancet, The</i> , 2018, 392, 1191-1192.	6.3	30
98	Mutations of the mitochondrial carrier translocase channel subunit TIM22 cause early-onset mitochondrial myopathy. <i>Human Molecular Genetics</i> , 2018, 27, 4135-4144.	1.4	30
99	The molecular pathology of pathogenic mitochondrial tRNA variants. <i>FEBS Letters</i> , 2021, 595, 1003-1024.	1.3	29
100	A proposed consensus panel of organisms for determining evolutionary conservation of mt-tRNA point mutations. <i>Mitochondrion</i> , 2012, 12, 533-538.	1.6	28
101	Long-term survival of neonatal mitochondrial complex III deficiency associated with a novel BCS1L gene mutation. <i>Molecular Genetics and Metabolism</i> , 2010, 100, 345-348.	0.5	27
102	Rapid-onset, linezolid-induced lactic acidosis in MELAS. <i>Mitochondrion</i> , 2011, 11, 992-993.	1.6	25
103	Scientific and Ethical Issues in Mitochondrial Donation. <i>New Bioethics</i> , 2018, 24, 57-73.	0.5	25
104	Resolving complexity in mitochondrial disease: Towards precision medicine. <i>Molecular Genetics and Metabolism</i> , 2019, 128, 19-29.	0.5	25
105	Mitochondrial Donation – Which Women Could Benefit?. <i>New England Journal of Medicine</i> , 2019, 380, 1971-1972.	13.9	25
106	Forecasting stroke-like episodes and outcomes in mitochondrial disease. <i>Brain</i> , 2022, 145, 542-554.	3.7	25
107	The genotypic and phenotypic spectrum of MTO1 deficiency. <i>Molecular Genetics and Metabolism</i> , 2018, 123, 28-42.	0.5	24
108	Outcome measures for children with mitochondrial disease: consensus recommendations for future studies from a Delphi-based international workshop. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 1267-1273.	1.7	24

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109	Pathogenic variants in <i>HTRA2</i> cause an early-onset mitochondrial syndrome associated with 3- $\alpha$ -methylglutaconic aciduria. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 121-130.	1.7	23
110	Clinical, biochemical, and genetic features associated with <i>VAR2</i> -related mitochondrial disease. <i>Human Mutation</i> , 2018, 39, 563-578.	1.1	22
111	The genetic basis of isolated mitochondrial complex II deficiency. <i>Molecular Genetics and Metabolism</i> , 2020, 131, 53-65.	0.5	22
112	Novel <i>MTND1</i> mutations cause isolated exercise intolerance, complex I deficiency and increased assembly factor expression. <i>Clinical Science</i> , 2015, 128, 895-904.	1.8	21
113	Loss-of-function mutations in <i>ISCA2</i> disrupt 4Fe-4S cluster machinery and cause a fatal leukodystrophy with hyperglycinemia and mtDNA depletion. <i>Human Mutation</i> , 2018, 39, 537-549.	1.1	21
114	A novel mitochondrial DNA tRNA <sup>Ala</sup> (A4267G) mutation in a sporadic patient with mitochondrial myopathy. <i>Neuromuscular Disorders</i> , 2002, 12, 659-664.	0.3	20
115	Maternally inherited mitochondrial DNA disease in consanguineous families. <i>European Journal of Human Genetics</i> , 2011, 19, 1226-1229.	1.4	20
116	Using a quantitative quadruple immunofluorescent assay to diagnose isolated mitochondrial Complex I deficiency. <i>Scientific Reports</i> , 2017, 7, 15676.	1.6	20
117	Dissecting the neuronal vulnerability underpinning Alpers <sup>TM</sup> syndrome: a clinical and neuropathological study. <i>Brain Pathology</i> , 2019, 29, 97-113.	2.1	20
118	Congenital myasthenic syndrome with mild intellectual disability caused by a recurrent <i>SLC25A1</i> variant. <i>European Journal of Human Genetics</i> , 2020, 28, 373-377.	1.4	20
119	Instability of the mitochondrial alanyl-tRNA synthetase underlies fatal infantile-onset cardiomyopathy. <i>Human Molecular Genetics</i> , 2019, 28, 258-268.	1.4	19
120	Height as a Clinical Biomarker of Disease Burden in Adult Mitochondrial Disease. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2019, 104, 2057-2066.	1.8	19
121	Measuring the effects of exercise in neuromuscular disorders: a systematic review and meta-analyses. <i>Wellcome Open Research</i> , 2020, 5, 84.	0.9	19
122	Childhood neurological presentation of a novel mitochondrial tRNA <sup>Val</sup> gene mutation. <i>Journal of the Neurological Sciences</i> , 2004, 225, 99-103.	0.3	18
123	Decreased male reproductive success in association with mitochondrial dysfunction. <i>European Journal of Human Genetics</i> , 2017, 25, 1162-1164.	1.4	18
124	Leigh syndrome caused by mutations in <i>MTFMT</i> is associated with a better prognosis. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 515-524.	1.7	17
125	Biallelic pathogenic variants in <i>NDUFC2</i> cause early-onset Leigh syndrome and stalled biogenesis of complex I. <i>EMBO Molecular Medicine</i> , 2020, 12, e12619.	3.3	17
126	Natural History of Leigh Syndrome: A Study of Disease Burden and Progression. <i>Annals of Neurology</i> , 2022, 91, 117-130.	2.8	17



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127	International Paediatric Mitochondrial Disease Scale. <i>Journal of Inherited Metabolic Disease</i> , 2016, 39, 705-712.	1.7	16
128	The adjunctive application of transcranial direct current stimulation in the management of de novo refractory epilepsy partialis continua in adolescent <i>onset POLG</i> -related mitochondrial disease. <i>Epilepsia Open</i> , 2018, 3, 103-108.	1.3	16
129	Sporadic myopathy and exercise intolerance associated with the mitochondrial 8328G>A tRNALys mutation. <i>Journal of Neurology</i> , 2007, 254, 1283-1285.	1.8	15
130	Compound heterozygous <i>RMND1</i> gene variants associated with chronic kidney disease, dilated cardiomyopathy and neurological involvement: a case report. <i>BMC Research Notes</i> , 2016, 9, 325.	0.6	15
131	Both mitochondrial DNA and mitonuclear gene mutations cause hearing loss through cochlear dysfunction. <i>Brain</i> , 2016, 139, e33-e33.	3.7	15
132	<i>CTBP1</i> -Arginine in Mitochondrial Encephalopathy, Lactic Acidosis, and Stroke-like Episodes. <i>Neurology</i> , 2022, 98, .	1.5	15
133	Mitochondrial respiratory chain disease in children undergoing cardiac transplantation: A prospective study. <i>International Journal of Cardiology</i> , 2012, 155, 305-306.	0.8	14
134	Long-term survival in a child with severe encephalopathy, multiple respiratory chain deficiency and <i>GFM1</i> mutations. <i>Frontiers in Genetics</i> , 2015, 6, 102.	1.1	13
135	<i>RMND1</i> -Related Leukoencephalopathy With Temporal Lobe Cysts and Hearing Loss—Another Mendelian Mimicker of Congenital Cytomegalovirus Infection. <i>Pediatric Neurology</i> , 2017, 66, 59-62.	1.0	12
136	Discrete gait characteristics are associated with m.3243A>G and m.8344A>G variants of mitochondrial disease and its pathological consequences. <i>Journal of Neurology</i> , 2014, 261, 73-82.	1.8	11
137	De novo <i>CTBP1</i> variant is associated with decreased mitochondrial respiratory chain activities. <i>Neurology: Genetics</i> , 2017, 3, e187.	0.9	11
138	Diabetes and Deafness: Is it sufficient to screen for the mitochondrial 3243A>G mutation alone?. <i>Diabetes Care</i> , 2007, 30, 2238-2239.	4.3	10
139	Mutations in the mitochondrial tRNASer(AGY) gene are associated with deafness, retinal degeneration, myopathy and epilepsy. <i>European Journal of Human Genetics</i> , 2012, 20, 897-904.	1.4	10
140	Novel <i>GFM2</i> variants associated with early-onset neurological presentations of mitochondrial disease and impaired expression of OXPHOS subunits. <i>Neurogenetics</i> , 2017, 18, 227-235.	0.7	10
141	Cognitive deficits in adult m.3243A>G and m.8344A>G related mitochondrial disease: importance of correcting for baseline intellectual ability. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 826-836.	1.7	10
142	<i>SURF1</i> related Leigh syndrome: Clinical and molecular findings of 16 patients from Turkey. <i>Molecular Genetics and Metabolism Reports</i> , 2020, 25, 100657.	0.4	10
143	Neuromuscular Junction Abnormalities in Mitochondrial Disease. <i>Neurology: Clinical Practice</i> , 2021, 11, 97-104.	0.8	10
144	Cerebral folate deficiency--mishaps and misdirection. <i>Brain</i> , 2012, 135, 2002-2003.	3.7	9

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145	Toward a mtDNA locus-specific mutation database using the LOVD platform. <i>Human Mutation</i> , 2012, 33, 1352-1358.	1.1	8
146	Prevalence and severity of voice and swallowing difficulties in mitochondrial disease. <i>International Journal of Language and Communication Disorders</i> , 2012, 47, 106-111.	0.7	8
147	Preliminary Evaluation of Clinician Rated Outcome Measures in Mitochondrial Disease. <i>Journal of Neuromuscular Diseases</i> , 2015, 2, 151-155.	1.1	8
148	The presence of anaemia negatively influences survival in patients with POLG disease. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 861-866.	1.7	8
149	A novel, pathogenic dinucleotide deletion in the mitochondrial MT-TY gene causing myasthenia-like features. <i>Neuromuscular Disorders</i> , 2020, 30, 661-668.	0.3	8
150	Early-onset coenzyme Q10 deficiency associated with ataxia and respiratory chain dysfunction due to novel pathogenic <i>COQ8A</i> variants, including a large intragenic deletion. <i>JIMD Reports</i> , 2020, 54, 45-53.	0.7	8
151	PPA2-associated sudden cardiac death: extending the clinical and allelic spectrum in 20 new families. <i>Genetics in Medicine</i> , 2021, 23, 2415-2425.	1.1	8
152	The Effect of tRNA[Ser]Sec Isopentenylation on Selenoprotein Expression. <i>International Journal of Molecular Sciences</i> , 2021, 22, 11454.	1.8	8
153	Delineating selective vulnerability of inhibitory interneurons in Alpers' syndrome. <i>Neuropathology and Applied Neurobiology</i> , 2022, 48, .	1.8	8
154	The pathogenic m.3243A>T mitochondrial DNA mutation is associated with a variable neurological phenotype. <i>Neuromuscular Disorders</i> , 2010, 20, 403-406.	0.3	7
155	The m.3291T>C mt-tRNA <sup>Leu</sup> (UUR) mutation is definitely pathogenic and causes multisystem mitochondrial disease. <i>Journal of the Neurological Sciences</i> , 2013, 325, 165-169.	0.3	7
156	Surveillance for variant CJD: should more children with neurodegenerative diseases have autopsies?. <i>Archives of Disease in Childhood</i> , 2019, 104, 360-365.	1.0	7
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