Rita Horvath

List of Publications by Year
in descending order

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299 papers	15,735 citations	16411 64 h-index	24915 109 g-index
310 all docs	310 docs citations	310 times ranked	16690 citing authors

#	Article	IF	Citations
1	Prevalence of nuclear and mitochondrial <scp>DNA</scp> mutations related to adult mitochondrial disease. Annals of Neurology, 2015, 77, 753-759.	2.8	706
2	Phenotypic spectrum associated with mutations of the mitochondrial polymerase Â gene. Brain, 2006, 129, 1674-1684.	3.7	397
3	Multi-system neurological disease is common in patients with OPA1 mutations. Brain, 2010, 133, 771-786.	3.7	385
4	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care â€" Preliminary Report. New England Journal of Medicine, 2021, 385, 1868-1880.	13.9	352
5	Universal heteroplasmy of human mitochondrial DNA. Human Molecular Genetics, 2013, 22, 384-390.	1.4	344
6	Whole-genome sequencing of patients with rare diseases in a national health system. Nature, 2020, 583, 96-102.	13.7	338
7	Clinical Expression of Leber Hereditary Optic Neuropathy Is Affected by the Mitochondrial DNAâ€"Haplogroup Background. American Journal of Human Genetics, 2007, 81, 228-233.	2.6	331
8	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
9	The myopathic form of coenzyme Q10 deficiency is caused by mutations in the electron-transferring-flavoprotein dehydrogenase (ETFDH) gene. Brain, 2007, 130, 2037-2044.	3.7	298
10	Mutation in TACO1, encoding a translational activator of COX I, results in cytochrome c oxidase deficiency and late-onset Leigh syndrome. Nature Genetics, 2009, 41, 833-837.	9.4	260
11	SDHAF1, encoding a LYR complex-II specific assembly factor, is mutated in SDH-defective infantile leukoencephalopathy. Nature Genetics, 2009, 41, 654-656.	9.4	233
12	Mutations in COX10 result in a defect in mitochondrial heme A biosynthesis and account for multiple, early-onset clinical phenotypes associated with isolated COX deficiency. Human Molecular Genetics, 2003, 12, 2693-2702.	1.4	219
13	Polymerase Î³ Gene POLG determines the risk of sodium valproate-induced liver toxicity. Hepatology, 2010, 52, 1791-1796.	3.6	219
14	Late onset Pompe disease: Clinical and neurophysiological spectrum of 38 patients including long-term follow-up in 18 patients. Neuromuscular Disorders, 2007, 17, 698-706.	0.3	208
15	Risk of developing a mitochondrial DNA deletion disorder. Lancet, The, 2004, 364, 592-596.	6.3	201
16	Mitochondrial aging is accelerated by anti-retroviral therapy through the clonal expansion of mtDNA mutations. Nature Genetics, 2011, 43, 806-810.	9.4	201
17	Extending the KCNQ2 encephalopathy spectrum. Neurology, 2013, 81, 1697-1703.	1.5	198
18	The Human Cytochrome c Oxidase Assembly Factors SCO1 and SCO2 Have Regulatory Roles in the Maintenance of Cellular Copper Homeostasis. Cell Metabolism, 2007, 5, 9-20.	7.2	197

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19	Identification of an X-Chromosomal Locus and Haplotype Modulating the Phenotype of a Mitochondrial DNA Disorder. American Journal of Human Genetics, 2005, 77, 1086-1091.	2.6	181
20	Patient care standards for primary mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. Genetics in Medicine, 2017, 19, 1380-1397.	1.1	173
21	The Prevalence and Natural History of Dominant Optic Atrophy Due to OPA1 Mutations. Ophthalmology, 2010, 117, 1538-1546.e1.	2.5	162
22	Mitochondria: Impaired mitochondrial translation in human disease. International Journal of Biochemistry and Cell Biology, 2014, 48, 77-84.	1.2	158
23	New treatments for mitochondrial diseaseâ€"no time to drop our standards. Nature Reviews Neurology, 2013, 9, 474-481.	4.9	157
24	Mutations in the SPG7 gene cause chronic progressive external ophthalmoplegia through disordered mitochondrial DNA maintenance. Brain, 2014, 137, 1323-1336.	3.7	151
25	Escobar Syndrome Is a Prenatal Myasthenia Caused by Disruption of the Acetylcholine Receptor Fetal Î³ Subunit. American Journal of Human Genetics, 2006, 79, 303-312.	2.6	146
26	Treatable childhood neuronopathy caused by mutations in riboflavin transporter RFVT2. Brain, 2014, 137, 44-56.	3.7	143
27	Mitochondrial Phosphateâ€"Carrier Deficiency: A Novel Disorder of Oxidative Phosphorylation. American Journal of Human Genetics, 2007, 80, 478-484.	2.6	142
28	The Cloning and Expression of a Human Creatine Transporter. Biochemical and Biophysical Research Communications, 1994, 204, 419-427.	1.0	136
29	Store-Operated Ca 2+ Entry Controls Induction of Lipolysis and the Transcriptional Reprogramming to Lipid Metabolism. Cell Metabolism, 2017, 25, 698-712.	7.2	131
30	EXOSC8 mutations alter mRNA metabolism and cause hypomyelination with spinal muscular atrophy and cerebellar hypoplasia. Nature Communications, 2014, 5, 4287.	5.8	120
31	Exome sequencing in undiagnosed inherited and sporadic ataxias. Brain, 2015, 138, 276-283.	3.7	120
32	Riboflavin-Responsive and -Non-responsive Mutations in FAD Synthase Cause Multiple Acyl-CoA Dehydrogenase and Combined Respiratory-Chain Deficiency. American Journal of Human Genetics, 2016, 98, 1130-1145.	2.6	118
33	Leigh syndrome caused by mutations in the flavoprotein (Fp) subunit of succinate dehydrogenase (SDHA). Journal of Neurology, Neurosurgery and Psychiatry, 2006, 77, 74-76.	0.9	115
34	Titin mutation segregates with hereditary myopathy with early respiratory failure. Brain, 2012, 135, 1695-1713.	3.7	113
35	Cytochrome c oxidase deficiency due to mutations in SCO2, encoding a mitochondrial copper-binding protein, is rescued by copper in human myoblasts. Human Molecular Genetics, 2001, 10, 3025-3035.	1.4	112
36	Molecular basis of infantile reversible cytochrome c oxidase deficiency myopathy. Brain, 2009, 132, 3165-3174.	3.7	112

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37	LPIN1 gene mutations: a major cause of severe rhabdomyolysis in early childhood. Human Mutation, 2010, 31, E1564-E1573.	1.1	112
38	Coenzyme Q10 deficiency and isolated myopathy. Neurology, 2006, 66, 253-255.	1.5	109
39	Safety and efficacy of deferiprone for pantothenate kinase-associated neurodegeneration: a randomised, double-blind, controlled trial and an open-label extension study. Lancet Neurology, The, 2019, 18, 631-642.	4.9	102
40	Hepatocerebral Mitochondrial DNA Depletion Syndrome Caused by Deoxyguanosine Kinase (DGUOK) Mutations. Archives of Neurology, 2006, 63, 1129.	4.9	101
41	Adult-onset cerebellar ataxia due to mutations inCABC1/ADCK3. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 174-178.	0.9	99
42	Absence of the Autophagy Adaptor SQSTM1/p62 Causes Childhood-Onset Neurodegeneration with Ataxia, Dystonia, and Gaze Palsy. American Journal of Human Genetics, 2016, 99, 735-743.	2.6	99
43	Neuropathology of white matter disease in Leber's hereditary optic neuropathy. Brain, 2004, 128, 35-41.	3.7	96
44	Functional defects due to spacer-region mutations of human mitochondrial DNA polymerase in a family with an ataxia-myopathy syndrome. Human Molecular Genetics, 2005, 14, 1907-1920.	1.4	96
45	Synaptotagmin 2 Mutations Cause an Autosomal-Dominant Form of Lambert-Eaton Myasthenic Syndrome and Nonprogressive Motor Neuropathy. American Journal of Human Genetics, 2014, 95, 332-339.	2.6	96
46	OPA1 mutations cause cytochrome c oxidase deficiency due to loss of wild-type mtDNA molecules. Human Molecular Genetics, 2010, 19, 3043-3052.	1.4	95
47	The phenotypic spectrum of neutral lipid storage myopathy due to mutations in the PNPLA2 gene. Journal of Neurology, 2011, 258, 1987-1997.	1.8	87
48	SPG7 mutations are a common cause of undiagnosed ataxia. Neurology, 2015, 84, 1174-1176.	1.5	87
49	Homozygous deletion in MICU1 presenting with fatigue and lethargy in childhood. Neurology: Genetics, 2016, 2, e59.	0.9	86
50	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. Brain, 2017, 140, 1561-1578.	3.7	85
51	The clinical, histochemical, and molecular spectrum of PEO1 (Twinkle)-linked adPEO. Neurology, 2010, 74, 1619-1626.	1.5	84
52	Frequency of mutations in the genes associated with hereditary sensory and autonomic neuropathy in a UK cohort. Journal of Neurology, 2012, 259, 1673-1685.	1.8	82
53	Homozygosity (E140K) in SCO2 causes delayed infantile onset of cardiomyopathy and neuropathy. Neurology, 2001, 57, 1440-1446.	1.5	81
54	Genetic heterogeneity of motor neuropathies. Neurology, 2017, 88, 1226-1234.	1.5	81

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55	Mutation screening of 75 candidate genes in 152 complex I deficiency cases identifies pathogenic variants in 16 genes includingNDUFB9. Journal of Medical Genetics, 2012, 49, 83-89.	1.5	78
56	Truncating and Missense Mutations in IGHMBP2 Cause Charcot-Marie Tooth Disease Type 2. American Journal of Human Genetics, 2014, 95, 590-601.	2.6	75
57	ANO10 mutations cause ataxia and coenzyme Q10 deficiency. Journal of Neurology, 2014, 261, 2192-2198.	1.8	74
58	Mitochondrial Diseases: A Diagnostic Revolution. Trends in Genetics, 2020, 36, 702-717.	2.9	73
59	Mitochondrial DNA transcription and translation: clinical syndromes. Essays in Biochemistry, 2018, 62, 321-340.	2.1	72
60	MFN2 mutations in Charcotâ€“Marieâ€“Tooth disease alter mitochondria-associated ER membrane function but do not impair bioenergetics. Human Molecular Genetics, 2019, 28, 1782-1800.	1.4	72
61	Titin founder mutation is a common cause of myofibrillar myopathy with early respiratory failure. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 331-338.	0.9	71
62	Adults with RRM2B-related mitochondrial disease have distinct clinical and molecular characteristics. Brain, 2012, 135, 3392-3403.	3.7	70
63	Parkinson syndrome, neuropathy, and myopathy caused by the mutation A8344G (MERRF) in tRNALys. Neurology, 2007, 68, 56-58.	1.5	69
64	Mutations in the Mitochondrial Citrate Carrier SLC25A1 are Associated with Impaired Neuromuscular Transmission. Journal of Neuromuscular Diseases, 2014, 1, 75-90.	1.1	69
65	The role of <scp>tRNA</scp> synthetases in neurological and neuromuscular disorders. FEBS Letters, 2018, 592, 703-717.	1.3	68
66	Dysregulation of Mitochondrial Ca2+ Uptake and Sarcolemma Repair Underlie Muscle Weakness and Wasting in Patients and Mice Lacking MICU1. Cell Reports, 2019, 29, 1274-1286.e6.	2.9	68
67	Abnormal neurological features predict poor survival and should preclude liver transplantation in patients with deoxyguanosine kinase deficiency. Liver Transplantation, 2008, 14, 1480-1485.	1.3	67
68	Nuclear factors involved in mitochondrial translation cause a subgroup of combined respiratory chain deficiency. Brain, 2011, 134, 183-195.	3.7	66
69	Update on clinical aspects and treatment of selected vitaminâ€responsive disorders II (riboflavin and) Tj ETQq1 1 0.784314 rgBT /Ove	1.7	66
70	Consensus-based statements for the management of mitochondrial stroke-like episodes. Wellcome Open Research, 2019, 4, 201.	0.9	66
71	Deficiency of the mitochondrial phosphate carrier presenting as myopathy and cardiomyopathy in a family with three affected children. Neuromuscular Disorders, 2011, 21, 803-808.	0.3	65
72	Variants in EXOSC9 Disrupt the RNA Exosome and Result in Cerebellar Atrophy with Spinal Motor Neuronopathy. American Journal of Human Genetics, 2018, 102, 858-873.	2.6	65

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73	Acute liver failure with subsequent cirrhosis as the primary manifestation of TRMU mutations. Journal of Inherited Metabolic Disease, 2011, 34, 197-201.	1.7	64
74	Accurate mitochondrial DNA sequencing using off-target reads provides a single test to identify pathogenic point mutations. Genetics in Medicine, 2014, 16, 962-971.	1.1	64
75	Clinical, pathological and functional characterization of riboflavin-responsive neuropathy. Brain, 2017, 140, 2820-2837.	3.7	64
76	The integrated stress response contributes to tRNA synthetaseâ€“associated peripheral neuropathy. Science, 2021, 373, 1156-1161.	6.0	64
77	Epilepsy in adults with mitochondrial disease: A cohort study. Annals of Neurology, 2015, 78, 949-957.	2.8	62
78	Revisiting mitochondrial diagnostic criteria in the new era of genomics. Genetics in Medicine, 2018, 20, 444-451.	1.1	62
79	Adult-onset glycogen storage disease type 2: clinico-pathological phenotype revisited. Neuropathology and Applied Neurobiology, 2007, 33, 070615152525006-???.	1.8	61
80	RRM2B mutations are frequent in familial PEO with multiple mtDNA deletions. Neurology, 2011, 76, 2032-2034.	1.5	59
81	Emerging therapies for mitochondrial disorders. Brain, 2016, 139, 1633-1648.	3.7	59
82	International Workshop:. Neuromuscular Disorders, 2017, 27, 1126-1137.	0.3	58
83	Reversion of hypertrophic cardiomyopathy in a patient with deficiency of the mitochondrial copper binding protein Sco2: Is there a potential effect of copper?. Journal of Inherited Metabolic Disease, 2004, 27, 67-79.	1.7	57
84	The 2-thiouridylase function of the human MTU1 (TRMU) enzyme is dispensable for mitochondrial translation. Human Molecular Genetics, 2011, 20, 4634-4643.	1.4	56
85	Mitochondrial DNA sequence characteristics modulate the size of the genetic bottleneck. Human Molecular Genetics, 2016, 25, 1031-1041.	1.4	53
86	Altered 2-thiouridylation impairs mitochondrial translation in reversible infantile respiratory chain deficiency. Human Molecular Genetics, 2013, 22, 4602-4615.	1.4	52
87	De Novo Mutations in EBF3 Cause a Neurodevelopmental Syndrome. American Journal of Human Genetics, 2017, 100, 138-150.	2.6	52
88	A novel mechanism causing imbalance of mitochondrial fusion and fission in human myopathies. Human Molecular Genetics, 2018, 27, 1186-1195.	1.4	52
89	Clinicoâ€Genetic, Imaging and Molecular Delineation of <scp>COQ8A</scp>â€Ataxia: A Multicenter Study of 59 Patients. Annals of Neurology, 2020, 88, 251-263.	2.8	52
90	First-line genomic diagnosis of mitochondrial disorders. Nature Reviews Genetics, 2018, 19, 399-400.	7.7	49

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91	Solve-RD: systematic pan-European data sharing and collaborative analysis to solve rare diseases. European Journal of Human Genetics, 2021, 29, 1325-1331.	1.4	49
92	Electrophysiologic features of SYT2 mutations causing a treatable neuromuscular syndrome. Neurology, 2015, 85, 1964-1971.	1.5	47
93	What is influencing the phenotype of the common homozygous polymerase-Î³ mutation p.Ala467Thr?. Brain, 2012, 135, 3614-3626.	3.7	46
94	Structural modeling of tissue-specific mitochondrial alanyl-tRNA synthetase (AARS2) defects predicts differential effects on aminoacylation. Frontiers in Genetics, 2015, 6, 21.	1.1	46
95	Metabolic effects of bezafibrate in mitochondrial disease. EMBO Molecular Medicine, 2020, 12, e11589.	3.3	45
96	Mutations in mtDNA-encoded cytochrome c oxidase subunit genes causing isolated myopathy or severe encephalomyopathy. Neuromuscular Disorders, 2005, 15, 851-857.	0.3	44
97	In vitro supplementation with dAMP/dGMP leads to partial restoration of mtDNA levels in mitochondrial depletion syndromes. Human Molecular Genetics, 2009, 18, 1590-1599.	1.4	44
98	Charcotâ€“Marieâ€“Tooth disease in Northern England: Figure 1. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 572-573.	0.9	44
99	Monitoring clinical progression with mitochondrial disease biomarkers. Brain, 2017, 140, 2530-2540.	3.7	44
100	Cross-sectional analysis of a large cohort with X-linked Charcot-Marie-Tooth disease (CMTX1). Neurology, 2017, 89, 927-935.	1.5	44
101	Clinical presentation and proteomic signature of patients with TANGO2 mutations. Journal of Inherited Metabolic Disease, 2020, 43, 297-308.	1.7	43
102	Adult-onset spinocerebellar ataxia syndromes due toMTATP6mutations. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 883-886.	0.9	42
103	Diagnosis of â€˜possibleâ€™ mitochondrial disease: an existential crisis. Journal of Medical Genetics, 2019, 56, 123-130.	1.5	42
104	Use of whole genome sequencing to determine genetic basis of suspected mitochondrial disorders: cohort study. BMJ, The, 2021, 375, e066288.	3.0	42
105	Collated mutations in mitochondrial DNA (mtDNA) depletion syndrome (excluding the mitochondrial) Tj ETQq1 1 0.784314 rgBT /Ove 1109-1112.	1.8	41
106	MFN2 mutations cause compensatory mitochondrial DNA proliferation. Brain, 2012, 135, e219-e219.	3.7	41
107	A new phenotype of brain iron accumulation with dystonia, optic atrophy, and peripheral neuropathy. Movement Disorders, 2012, 27, 789-793.	2.2	41
108	Clonal Expansion of Secondary Mitochondrial DNA Deletions Associated With Spinocerebellar Ataxia Type 28. JAMA Neurology, 2015, 72, 106.	4.5	41

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109	Valproic acid triggers increased mitochondrial biogenesis in POLG-deficient fibroblasts. Molecular Genetics and Metabolism, 2014, 112, 57-63.	0.5	40
110	Late-onset ptosis and myopathy in a patient with a heterozygous insertion in POLG2. Journal of Neurology, 2010, 257, 1517-1523.	1.8	39
111	SCP2 mutations and neurodegeneration with brain iron accumulation. Neurology, 2015, 85, 1909-1911.	1.5	39
112	Natural history of Charcot-Marie-Tooth disease type 2A: a large international multicentre study. Brain, 2020, 143, 3589-3602.	3.7	39
113	Gentamicin treatment in McArdle disease: Failure to correct myophosphorylase deficiency. Neurology, 2006, 66, 285-286.	1.5	38
114	Mitochondrial myopathies: developments in treatment. Current Opinion in Neurology, 2010, 23, 459-465.	1.8	38
115	Multisystem fatal infantile disease caused by a novel homozygous EARS2 mutation. Brain, 2013, 136, e228-e228.	3.7	38
116	Identification of rare DNA variants in mitochondrial disorders with improved array-based sequencing. Nucleic Acids Research, 2011, 39, 44-58.	6.5	37
117	Mitochondrial pathology in progressive cerebellar ataxia. Cerebellum and Ataxias, 2015, 2, 16.	1.9	37
118	Reversible infantile mitochondrial diseases. Journal of Inherited Metabolic Disease, 2015, 38, 427-435.	1.7	37
119	Characterization of Human SCO1 and COX17 Genes in Mitochondrial Cytochrome-c-Oxidase Deficiency. Biochemical and Biophysical Research Communications, 2000, 276, 530-533.	1.0	36
120	Advances in the diagnosis of inherited neuromuscular diseases and implications for therapy development. Lancet Neurology, The, 2020, 19, 522-532.	4.9	36
121	Leber's hereditary optic neuropathy presenting as multiple sclerosis-like disease of the CNS. Journal of Neurology, 2000, 247, 65-67.	1.8	35
122	Severe nemaline myopathy caused by mutations of the stop codon of the skeletal muscle alpha actin gene (ACTA1). Neuromuscular Disorders, 2006, 16, 541-547.	0.3	35
123	Heteroplasmic mutation in the anticodon-stem of mitochondrial tRNAVal causing MNGIE-like gastrointestinal dysmotility and cachexia. Journal of Neurology, 2009, 256, 810-815.	1.8	35
124	Infantile Encephaloneuromyopathy and Defective Mitochondrial Translation Are Due to a Homozygous RMND1 Mutation. American Journal of Human Genetics, 2012, 91, 729-736.	2.6	35
125	Altered RNA metabolism due to a homozygousRBM7mutation in a patient with spinal motor neuropathy. Human Molecular Genetics, 2016, 25, ddw149.	1.4	35
126	Childhood onset mitochondrial myopathy and lactic acidosis caused by a stop mutation in the mitochondrial cytochrome c oxidase III gene. Journal of Medical Genetics, 2002, 39, 812-816.	1.5	34

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127	A novel mitochondrial MTND5 frameshift mutation causing isolated complex I deficiency, renal failure and myopathy. Neuromuscular Disorders, 2010, 20, 131-135.	0.3	34
128	A novel de novo STXBP1 mutation is associated with mitochondrial complex I deficiency and late-onset juvenile-onset parkinsonism. Neurogenetics, 2015, 16, 65-67.	0.7	34
129	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. European Journal of Human Genetics, 2021, 29, 1337-1347.	1.4	34
130	How Can We Treat Mitochondrial Encephalomyopathies? Approaches to Therapy. Neurotherapeutics, 2008, 5, 558-568.	2.1	33
131	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.	1.8	33
132	Mitochondrial DNA deletions in muscle satellite cells: implications for therapies. Human Molecular Genetics, 2013, 22, 4739-4747.	1.4	33
133	Mitochondrial dysfunction in liver failure requiring transplantation. Journal of Inherited Metabolic Disease, 2016, 39, 427-436.	1.7	33
134	Intersection of Proteomics and Genomics to â€œSolve the Unsolvedâ€•in Rare Disorders such as Neurodegenerative and Neuromuscular Diseases. Proteomics - Clinical Applications, 2018, 12, 1700073.	0.8	33
135	Pathogenic variants in MTâ€ATP6: A United Kingdomâ€“based mitochondrial disease cohort study. Annals of Neurology, 2019, 86, 310-315.	2.8	33
136	De Novo and Bi-allelic Pathogenic Variants in NARS1 Cause Neurodevelopmental Delay Due to Toxic Gain-of-Function and Partial Loss-of-Function Effects. American Journal of Human Genetics, 2020, 107, 311-324.	2.6	32
137	AAV9-mediated Schwann cell-targeted gene therapy rescues a model of demyelinating neuropathy. Gene Therapy, 2021, 28, 659-675.	2.3	32
138	Clinical and neuropathological findings in patients with TACO1 mutations. Neuromuscular Disorders, 2010, 20, 720-724.	0.3	31
139	In vitro supplementation with deoxynucleoside monophosphates rescues mitochondrial DNA depletion. Molecular Genetics and Metabolism, 2012, 107, 95-103.	0.5	31
140	Genotype/phenotype correlations in AARS-related neuropathy in a cohort of patients from the United Kingdom and Ireland. Journal of Neurology, 2015, 262, 1899-1908.	1.8	31
141	Mitochondrial oxodicarboxylate carrier deficiency is associated with mitochondrial DNA depletion and spinal muscular atrophyâ€“like disease. Genetics in Medicine, 2018, 20, 1224-1235.	1.1	31
142	Mutations inELAC2associated with hypertrophic cardiomyopathy impair mitochondrial tRNA 3â€²â€end processing. Human Mutation, 2019, 40, 1731-1748.	1.1	31
143	Fatal Neonatal-Onset Mitochondrial Respiratory Chain Disease with T Cell Immunodeficiency. Pediatric Research, 2006, 60, 321-326.	1.1	30
144	OPA1 IN MULTIPLE MITOCHONDRIAL DNA DELETION DISORDERS. Neurology, 2008, 71, 1829-1831.	1.5	30

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145	AMACR mutations cause late-onset autosomal recessive cerebellar ataxia. Neurology, 2011, 76, 1768-1770.	1.5	30
146	Two recurrent mutations are associated with GNE myopathy in the North of Britain. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 1359-1365.	0.9	30
147	alpha-Tocopherol/lipid ratio in blood is decreased in patients with Leber's hereditary optic neuropathy and asymptomatic carriers of the 11778 mtDNA mutation. Journal of Neurology, Neurosurgery and Psychiatry, 2001, 70, 359-362.	0.9	29
148	SACS variants are a relevant cause of autosomal recessive hereditary motor and sensory neuropathy. Human Genetics, 2018, 137, 911-919.	1.8	29
149	Salbutamol modifies the neuromuscular junction in a mouse model of ColQ myasthenic syndrome. Human Molecular Genetics, 2019, 28, 2339-2351.	1.4	29
150	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
151	Phenotypic variability of TRPV4 related neuropathies. Neuromuscular Disorders, 2015, 25, 516-521.	0.3	27
152	Demyelinating disease of central and peripheral nervous systems associated with a A8344G mutation in tRNALys. Neuromuscular Disorders, 2009, 19, 275-278.	0.3	26
153	Prevalence of neurogenetic disorders in the North of England. Neurology, 2015, 85, 1195-1201.	1.5	26
154	Mutations in glycyl-tRNA synthetase impair mitochondrial metabolism in neurons. Human Molecular Genetics, 2018, 27, 2187-2204.	1.4	26
155	Metabolic shift underlies recovery in reversible infantile respiratory chain deficiency. EMBO Journal, 2020, 39, e105364.	3.5	26
156	Clinical and functional characterisation of the combined respiratory chain defect in two sisters due to autosomal recessive mutations in MTFMT. Mitochondrion, 2013, 13, 743-748.	1.6	25
157	Abnormal retinal thickening is a common feature among patients with ARSACS-related phenotypes: TableÂ1. British Journal of Ophthalmology, 2014, 98, 711-713.	2.1	25
158	Two families with autosomal dominant progressive external ophthalmoplegia. Journal of Neurology, Neurosurgery and Psychiatry, 2004, 75, 1125-1128.	0.9	24
159	Novel HSPB1 mutation causes both motor neuronopathy and distal myopathy. Neurology: Genetics, 2016, 2, e110.	0.9	24
160	Pathogenic mtDNA mutations causing mitochondrial myopathy: The need for muscle biopsy. Neurology: Genetics, 2016, 2, e82.	0.9	24
161	The genotypic and phenotypic spectrum of MTO1 deficiency. Molecular Genetics and Metabolism, 2018, 123, 28-42.	0.5	24
162	Neuromuscular Junction Changes in a Mouse Model of Charcot-Marie-Tooth Disease Type 4C. International Journal of Molecular Sciences, 2018, 19, 4072.	1.8	24

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163	Predicting the contribution of novel POLG mutations to human disease through analysis in yeast model. Mitochondrion, 2011, 11, 182-190.	1.6	23
164	Late-onset respiratory failure due to TK2 mutations causing multiple mtDNA deletions. Neurology, 2013, 81, 2051-2053.	1.5	23
165	Respiratory chain deficiency in nonmitochondrial disease. Neurology: Genetics, 2015, 1, e6.	0.9	23
166	Congenital cataract, muscular hypotonia, developmental delay and sensorineural hearing loss associated with a defect in copper metabolism. Journal of Inherited Metabolic Disease, 2005, 28, 479-492.	1.7	22
167	Novel genetic and neuropathological insights in neurogenic muscle weakness, ataxia, and retinitis pigmentosa (NARP). Muscle and Nerve, 2016, 54, 328-333.	1.0	22
168	Novel homozygous RARS2 mutation in two siblings without pontocerebellar hypoplasia â€“ further expansion of the phenotypic spectrum. Orphanet Journal of Rare Diseases, 2016, 11, 140.	1.2	22
169	Prominent Sensorimotor Neuropathy Due to SACS Mutations Revealed by Whole-Exome Sequencing. Archives of Neurology, 2012, 69, 1351-4.	4.9	21
170	Progressive Brain Iron Accumulation in Neuroferritinopathy Measured by the Thalamic T2* Relaxation Rate. American Journal of Neuroradiology, 2012, 33, 1810-1813.	1.2	21
171	Clonal expansion of mtDNA deletions: different disease models assessed by digital droplet PCR in single muscle cells. Scientific Reports, 2018, 8, 11682.	1.6	21
172	A guide to writing systematic reviews of rare disease treatments to generate FAIR-compliant datasets: building a Treatabolome. Orphanet Journal of Rare Diseases, 2020, 15, 206.	1.2	21
173	The role of complex I genes in MELAS: A novel heteroplasmic mutation 3380G>A in ND1 of mtDNA. Neuromuscular Disorders, 2008, 18, 553-556.	0.3	20
174	Behr's Syndrome is Typically Associated with Disturbed Mitochondrial Translation and Mutations in the C12orf65 Gene. Journal of Neuromuscular Diseases, 2014, 1, 55-63.	1.1	20
175	Frequency of rare recessive mutations in unexplained late onset cerebellar ataxia. Journal of Neurology, 2015, 262, 1822-1827.	1.8	20
176	Biomarkers predict outcome in Charcot-Marie-Tooth disease 1A. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 941-952.	0.9	20
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