Rita Horvath

List of Publications by Year
in descending order

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299	15,735	18887 64	28425 109
papers	citations	h-index	g-index

310	310	310	17978
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Elucidating the molecular mechanisms associated with TARS2-related mitochondrial disease. Human Molecular Genetics, 2022, 31, 523-534.	1.4	12
2	A de novo <scp>CSDE1</scp> variant causing neurodevelopmental delay, intellectual disability, neurologic and psychiatric symptoms in a child of consanguineous parents. American Journal of Medical Genetics, Part A, 2022, 188, 283-291.	0.7	1
3	High diagnostic rate of trio exome sequencing in consanguineous families with neurogenetic diseases. Brain, 2022, 145, 1507-1518.	3.7	14
4	Novel insights into PORCN mutations, associated phenotypes and pathophysiological aspects. Orphanet Journal of Rare Diseases, 2022, 17, 29.	1.2	3
5	Identification of a novel homozygous synthesis of cytochrome c oxidase 2 variant in siblings with earlyâ€onset axonal Charcotâ€Marieâ€Tooth disease. Human Mutation, 2022, 43, 477-486.	1.1	3
6	NCAM1 and GDF15 are biomarkers of Charcot-Marie-Tooth disease in patients and mice. Brain, 2022, 145, 3999-4015.	3.7	12
7	The RDâ€Connect Genomeâ€Phenome Analysis Platform: Accelerating diagnosis, research, and gene discovery for rare diseases. Human Mutation, 2022, , .	1.1	18
8	Biallelic ADAM22 pathogenic variants cause progressive encephalopathy and infantile-onset refractory epilepsy. Brain, 2022, 145, 2301-2312.	3.7	8
9	Molecular and neurological features of MELAS syndrome in paediatric patients: A case series and review of the literature. Molecular Genetics & Genomic Medicine, 2022, 10, e1955.	0.6	8
10	A translatable RNAi-driven gene therapy silences PMP22/Pmp22 genes and improves neuropathy in CMT1A mice. Journal of Clinical Investigation, 2022, 132, .	3.9	18
11	Targeted Therapies for Hereditary Peripheral Neuropathies: Systematic Review and Steps Towards a â€˜treatabolomeâ€™. Journal of Neuromuscular Diseases, 2021, 8, 383-400.	1.1	10
12	Inherited neuropathies with predominant upper limb involvement: genetic heterogeneity and overlapping pathologies. European Journal of Neurology, 2021, 28, 297-304.	1.7	4
13	Mitochondrial Translation Deficiencies. , 2021, , 95-117.		0
14	Molecular pathophysiology of human MICU1 deficiency. Neuropathology and Applied Neurobiology, 2021, 47, 840-855.	1.8	15
15	Results from a 3-year Non-interventional, Observational Disease Monitoring Program in Adults with GNE Myopathy. Journal of Neuromuscular Diseases, 2021, 8, 225-234.	1.1	9
16	CMT2N-causing aminoacylation domain mutants enable Nrp1 interaction with AlaRS. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	16
17	AAV9-mediated Schwann cell-targeted gene therapy rescues a model of demyelinating neuropathy. Gene Therapy, 2021, 28, 659-675.	2.3	32
18	INPP5K and SIL1 associated pathologies with overlapping clinical phenotypes converge through dysregulation of PHGDH. Brain, 2021, 144, 2427-2442.	3.7	7

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19	[11C]PK11195-PET Brain Imaging of the Mitochondrial Translocator Protein in Mitochondrial Disease. Neurology, 2021, 96, e2761-e2773.	1.5	7
20	Muscle fat replacement and modified ragged red fibers in two patients with reversible infantile respiratory chain deficiency. Neuromuscular Disorders, 2021, 31, 551-557.	0.3	2
21	Exome reanalysis and proteomic profiling identified TRIP4 as a novel cause of cerebellar hypoplasia and spinal muscular atrophy (PCH1). European Journal of Human Genetics, 2021, 29, 1348-1353.	1.4	10
22	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
23	A MT-TL1 variant identified by whole exome sequencing in an individual with intellectual disability, epilepsy, and spastic tetraparesis. European Journal of Human Genetics, 2021, 29, 1359-1368.	1.4	7
24	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. European Journal of Human Genetics, 2021, 29, 1337-1347.	1.4	34
25	Targeted Therapies for Leigh Syndrome: Systematic Review and Steps Towards a â€˜Treatabolomeâ€™. Journal of Neuromuscular Diseases, 2021, 8, 885-897.	1.1	6
26	MitoPhen database: a human phenotype ontology-based approach to identify mitochondrial DNA diseases. Nucleic Acids Research, 2021, 49, 9686-9695.	6.5	14
27	Modelling Charcot-Marie-Tooth disease in a dish reveals common cell type-specific alterations. Brain, 2021, 144, 2234-2236.	3.7	0
28	The integrated stress response contributes to tRNA synthetaseâ€“associated peripheral neuropathy. Science, 2021, 373, 1156-1161.	6.0	64
29	NEW GENES AND DISEASES. Neuromuscular Disorders, 2021, 31, S143.	0.3	0
30	Autosomal recessive variants in TUBGCP2 alter the Î³-tubulin ring complex leading to neurodevelopmental disease. IScience, 2021, 24, 101948.	1.9	6
31	Bi-allelic variants in SPATA5L1 lead to intellectual disability, spastic-dystonic cerebral palsy, epilepsy, and hearing loss. American Journal of Human Genetics, 2021, 108, 2006-2016.	2.6	11
32	Intracellular Lipid Accumulation and Mitochondrial Dysfunction Accompanies Endoplasmic Reticulum Stress Caused by Loss of the Co-chaperone DNAJC3. Frontiers in Cell and Developmental Biology, 2021, 9, 710247.	1.8	13
33	Homozygous WASHC4 variant in two sisters causes a syndromic phenotype defined by dysmorphisms, intellectual disability, profound developmental disorder, and skeletal muscle involvement. Journal of Pathology, 2021, , .	2.1	5
34	Alpers- and MNGIE-like disease with disturbed CSF folate transport and an unusual mode of genetic transmission of POLG mutations: a case report. Journal of International Child Neurology Association, 2021, 1, .	0.0	1
35	Primary mitochondrial myopathies in childhood. Neuromuscular Disorders, 2021, 31, 978-987.	0.3	4
36	Use of whole genome sequencing to determine genetic basis of suspected mitochondrial disorders: cohort study. BMJ, The, 2021, 375, e066288.	3.0	42

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37	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
38	White Matter Hyperintensities and Cerebral Microbleeds in Ataxia-Telangiectasia. Neurology: Genetics, 2021, 7, e640.	0.9	2
39	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
40	Severe neurodevelopmental disease caused by a homozygous TLK2 variant. European Journal of Human Genetics, 2020, 28, 383-387.	1.4	6
41	Clinical presentation and proteomic signature of patients with TANGO2 mutations. Journal of Inherited Metabolic Disease, 2020, 43, 297-308.	1.7	43
42	Mitochondrial Diseases: A Diagnostic Revolution. Trends in Genetics, 2020, 36, 702-717.	2.9	73
43	Biallelic loss of function variants in <scp>SYT2</scp> cause a treatable congenital onset presynaptic myasthenic syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2272-2283.	0.7	20
44	Behr syndrome and hypertrophic cardiomyopathy in a family with a novel UCHL1 deletion. Journal of Neurology, 2020, 267, 3643-3649.	1.8	8
45	COL4A1 -related autosomal recessive encephalopathy in 2 Turkish children. Neurology: Genetics, 2020, 6, e392.	0.9	9
46	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
47	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
48	The genetic landscape of axonal neuropathies in the middle-aged and elderly. Neurology, 2020, 95, e3163-e3179.	1.5	19
49	Clinical and Genetic Features in a Series of Eight Unrelated Patients with Neuropathy Due to Glycyl-tRNA Synthetase (GARS) Variants. Journal of Neuromuscular Diseases, 2020, 7, 137-143.	1.1	16
50	Advances in the diagnosis of inherited neuromuscular diseases and implications for therapy development. Lancet Neurology, The, 2020, 19, 522-532.	4.9	36
51	Whole-genome sequencing of patients with rare diseases in a national health system. Nature, 2020, 583, 96-102.	13.7	338
52	Metabolic effects of bezafibrate in mitochondrial disease. EMBO Molecular Medicine, 2020, 12, e11589.	3.3	45
53	Improved Diagnosis of Rare Disease Patients through Systematic Detection of Runs of Homozygosity. Journal of Molecular Diagnostics, 2020, 22, 1205-1215.	1.2	14
54	Multiple acyl-coenzyme A dehydrogenase deficiency shows a possible founder effect and is the most frequent cause of lipid storage myopathy in Iran. Journal of the Neurological Sciences, 2020, 411, 116707.	0.3	14

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55	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
56	Clinico-Genetic, Imaging and Molecular Delineation of COQ8A-Ataxia: A Multicenter Study of 59 Patients. , 2020, 88, 251.		1
57	Natural history of Charcot-Marie-Tooth disease type 2A: a large international multicentre study. Brain, 2020, 143, 3589-3602.	3.7	39
58	Metabolic shift underlies recovery in reversible infantile respiratory chain deficiency. EMBO Journal, 2020, 39, e105364.	3.5	26
59	RNA exosome mutations in pontocerebellar hypoplasia alter ribosome biogenesis and p53 levels. Life Science Alliance, 2020, 3, e202000678.	1.3	17
60	Confirmation of TACO1 as a Leigh Syndrome Disease Gene in Two Additional Families. Journal of Neuromuscular Diseases, 2020, 7, 301-308.	1.1	8
61	Mitochondrial disorders due to mutations in the nuclear genome. , 2020, , 415-425.		0
62	Modulation of the Acetylcholine Receptor Clustering Pathway Improves Neuromuscular Junction Structure and Muscle Strength in a Mouse Model of Congenital Myasthenic Syndrome. Frontiers in Molecular Neuroscience, 2020, 13, 594220.	1.4	5
63	Identification of Cellular Pathogenicity Markers for SIL1 Mutations Linked to Marinesco-SjÃ¶gren Syndrome. Frontiers in Neurology, 2019, 10, 562.	1.1	5
64	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
65	Diagnosis of â€˜possibleâ€™ mitochondrial disease: an existential crisis. Journal of Medical Genetics, 2019, 56, 123-130.	1.5	42
66	HADHA and HADHB gene associated phenotypes - Identification of rare variants in a patient cohort by Next Generation Sequencing. Molecular and Cellular Probes, 2019, 44, 14-20.	0.9	20
67	Salbutamol modifies the neuromuscular junction in a mouse model of ColQ myasthenic syndrome. Human Molecular Genetics, 2019, 28, 2339-2351.	1.4	29
68	Identification of Candidate Protein Markers in Skeletal Muscle of Laminin-211-Deficient CMD Type 1A-Patients. Frontiers in Neurology, 2019, 10, 470.	1.1	14
69	Pathogenic variants in MTâ€ATP6: A United Kingdomâ€“based mitochondrial disease cohort study. Annals of Neurology, 2019, 86, 310-315.	2.8	33
70	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
71	Mutations inELAC2associated with hypertrophic cardiomyopathy impair mitochondrial tRNA 3â€²â€end processing. Human Mutation, 2019, 40, 1731-1748.	1.1	31
72	Mitochondrial Depletion Syndromes. , 2019, , 183-204.		0

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73	Diagnostic Approach to Mitochondrial Diseases. , 2019, , 281-287.		0
74	Nucleoside supplementation modulates mitochondrial DNA copy number in thedguok â^'/â^'zebrafish. Human Molecular Genetics, 2019, 28, 796-803.	1.4	14
75	SIL1 deficiency causes degenerative changes of peripheral nerves and neuromuscular junctions in fish, mice and human. Neurobiology of Disease, 2019, 124, 218-229.	2.1	7
76	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
77	Consensus-based statements for the management of mitochondrial stroke-like episodes. Wellcome Open Research, 2019, 4, 201.	0.9	66
78	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
79	Heterozygous SSBP1 start loss mutation co-segregates with hearing loss and the m.1555A>G mtDNA variant in a large multigenerational family. Brain, 2018, 141, 55-62.	3.7	19
80	Mutations in glycyl-tRNA synthetase impair mitochondrial metabolism in neurons. Human Molecular Genetics, 2018, 27, 2187-2204.	1.4	26
81	PFN2 and GAMT as common molecular determinants of axonal Charcot-Marie-Tooth disease. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 870-878.	0.9	16
82	A novel mechanism causing imbalance of mitochondrial fusion and fission in human myopathies. Human Molecular Genetics, 2018, 27, 1186-1195.	1.4	52
83	The role of <scp>tRNA</scp> synthetases in neurological and neuromuscular disorders. FEBS Letters, 2018, 592, 703-717.	1.3	68
84	Reply: POLR3A variants in hereditary spastic paraplegia and ataxia. Brain, 2018, 141, e2-e2.	3.7	10
85	Multifocal demyelinating motor neuropathy and hamartoma syndrome associated with a de novo PTEN mutation. Neurology, 2018, 90, e1842-e1848.	1.5	4
86	How to Spot Congenital Myasthenic Syndromes Resembling the Lambertâ€“Eaton Myasthenic Syndrome? A Brief Review of Clinical, Electrophysiological, and Genetics Features. NeuroMolecular Medicine, 2018, 20, 205-214.	1.8	4
87	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
88	Revisiting mitochondrial diagnostic criteria in the new era of genomics. Genetics in Medicine, 2018, 20, 444-451.	1.1	62
89	The genotypic and phenotypic spectrum of MTO1 deficiency. Molecular Genetics and Metabolism, 2018, 123, 28-42.	0.5	24
90	SACS variants are a relevant cause of autosomal recessive hereditary motor and sensory neuropathy. Human Genetics, 2018, 137, 911-919.	1.8	29

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91	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
92	First-line genomic diagnosis of mitochondrial disorders. Nature Reviews Genetics, 2018, 19, 399-400.	7.7	49
93	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
94	Mitochondrial DNA transcription and translation: clinical syndromes. Essays in Biochemistry, 2018, 62, 321-340.	2.1	72
95	Novel SBF2 mutations and clinical spectrum of Charcotâ€Marieâ€Tooth neuropathy type 4B2. Clinical Genetics, 2018, 94, 467-472.	1.0	7
96	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
97	An Unusual Retinal Phenotype Associated With a Mutation in Sterol Carrier Protein SCP2. JAMA Ophthalmology, 2017, 135, 167.	1.4	7
98	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
99	Genetic heterogeneity of motor neuropathies. Neurology, 2017, 88, 1226-1234.	1.5	81
100	Lifetime exercise intolerance with lactic acidosis as key manifestation of novel compound heterozygous ACAD9 mutations causing complex I deficiency. Neuromuscular Disorders, 2017, 27, 473-476.	0.3	10
101	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. Brain, 2017, 140, 1561-1578.	3.7	85
102	De Novo Mutations in EBF3 Cause a Neurodevelopmental Syndrome. American Journal of Human Genetics, 2017, 100, 138-150.	2.6	52
103	PLP1 mutations and central demyelination. Neurology: Clinical Practice, 2017, 7, 451-454.	0.8	0
104	Monitoring clinical progression with mitochondrial disease biomarkers. Brain, 2017, 140, 2530-2540.	3.7	44
105	Clinical, pathological and functional characterization of riboflavin-responsive neuropathy. Brain, 2017, 140, 2820-2837.	3.7	64
106	Biomarkers predict outcome in Charcot-Marie-Tooth disease 1A. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 941-952.	0.9	20
107	International Workshop:. Neuromuscular Disorders, 2017, 27, 1126-1137.	0.3	58
108	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

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109	Cross-sectional analysis of a large cohort with X-linked Charcot-Marie-Tooth disease (CMTX1). Neurology, 2017, 89, 927-935.	1.5	44
110	Response to Newman et al.. Genetics in Medicine, 2017, 19, 1380-1380.	1.1	3
111	The Effect of Neurological Genomics and Personalized Mitochondrial Medicine. JAMA Neurology, 2017, 74, 11.	4.5	2
112	Multidrug Resistant Pseudomonas Mycotic Pseudoaneurysm following Cardiac Transplant Bridged by Ventricular Assistant Device. Case Reports in Infectious Diseases, 2017, 2017, 1-4.	0.2	8
113	Drosophila studies support a role for a presynaptic synaptotagmin mutation in a human congenital myasthenic syndrome. PLoS ONE, 2017, 12, e0184817.	1.1	12
114	Novel HSPB1 mutation causes both motor neuronopathy and distal myopathy. Neurology: Genetics, 2016, 2, e110.	0.9	24
115	Novel genetic and neuropathological insights in neurogenic muscle weakness, ataxia, and retinitis pigmentosa (NARP). Muscle and Nerve, 2016, 54, 328-333.	1.0	22
116	Genetic analyses and clinical features in a series of eight unrelated patients with Glycyl-tRNA synthetase (GARS) variants. Neuromuscular Disorders, 2016, 26, S141.	0.3	1
117	Cysteine Supplementation May be Beneficial in a Subgroup of Mitochondrial Translation Deficiencies. Journal of Neuromuscular Diseases, 2016, 3, 363-379.	1.1	17
118	Homozygous deletion in MICU1 presenting with fatigue and lethargy in childhood. Neurology: Genetics, 2016, 2, e59.	0.9	86
119	A Mutation in the Flavin Adenine Dinucleotide-Dependent Oxidoreductase FOXRED1 Results in Cell-Type-Specific Assembly Defects in Oxidative Phosphorylation Complexes I and II. Molecular and Cellular Biology, 2016, 36, 2132-2140.	1.1	19
120	Altered RNA metabolism due to a homozygousRBM7mutation in a patient with spinal motor neuropathy. Human Molecular Genetics, 2016, 25, ddw149.	1.4	35
121	Emerging therapies for mitochondrial disorders. Brain, 2016, 139, 1633-1648.	3.7	59
122	Pathogenic mtDNA mutations causing mitochondrial myopathy: The need for muscle biopsy. Neurology: Genetics, 2016, 2, e82.	0.9	24
123	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
124	Phenotypic convergence of Menkes and Wilson disease. Neurology: Genetics, 2016, 2, e119.	0.9	18
125	The swinging pendulum of biomarkers in mitochondrial disease. Neurology, 2016, 87, 2286-2287.	1.5	5
126	Amyloidâ€î² in mitochondrial disease: mutation in a human metallopeptidase links amyloidotic neurodegeneration with mitochondrial processing. EMBO Molecular Medicine, 2016, 8, 173-175.	3.3	5

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127	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
128	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
129	Both mitochondrial DNA and mitonuclear gene mutations cause hearing loss through cochlear dysfunction. Brain, 2016, 139, e33-e33.	3.7	15
130	Mitochondrial dysfunction in liver failure requiring transplantation. Journal of Inherited Metabolic Disease, 2016, 39, 427-436.	1.7	33
131	Mitochondrial DNA sequence characteristics modulate the size of the genetic bottleneck. Human Molecular Genetics, 2016, 25, 1031-1041.	1.4	53
132	Reversible Infantile Respiratory Chain Deficiency. , 2016, , 127-133.		0
133	Metabolic stroke in childhood: Diagnostic approach and suggestions for therapy. Journal of Pediatric Neurology, 2015, 08, 321-332.	0.0	1
134	Epilepsy in adults with mitochondrial disease: A cohort study. Annals of Neurology, 2015, 78, 949-957.	2.8	62
135	Mitochondrial pathology in progressive cerebellar ataxia. Cerebellum and Ataxias, 2015, 2, 16.	1.9	37
136	Exosomal Protein Deficiencies: How Abnormal RNA Metabolism Results in Childhood-Onset Neurological Diseases. Journal of Neuromuscular Diseases, 2015, 2, S31-S37.	1.1	13
137	ATP Synthase Deficiency due to TMEM70 Mutation Leads to Ultrastructural Mitochondrial Degeneration and Is Amenable to Treatment. BioMed Research International, 2015, 2015, 1-10.	0.9	10
138	Nuclear-mitochondrial proteins: too much to process?: Figure 1. Brain, 2015, 138, 1451-1453.	3.7	2
139	Prevalence of nuclear and mitochondrial <scp>DNA</scp> mutations related to adult mitochondrial disease. Annals of Neurology, 2015, 77, 753-759.	2.8	706
140	Frequency of rare recessive mutations in unexplained late onset cerebellar ataxia. Journal of Neurology, 2015, 262, 1822-1827.	1.8	20
141	Clonal Expansion of Secondary Mitochondrial DNA Deletions Associated With Spinocerebellar Ataxia Type 28. JAMA Neurology, 2015, 72, 106.	4.5	41
142	Respiratory chain deficiency in nonmitochondrial disease. Neurology: Genetics, 2015, 1, e6.	0.9	23
143	Investigating the role of the physiological isoform switch of cytochrome c oxidase subunits in reversible mitochondrial disease. International Journal of Biochemistry and Cell Biology, 2015, 63, 32-40.	1.2	14
144	Reversible infantile mitochondrial diseases. Journal of Inherited Metabolic Disease, 2015, 38, 427-435.	1.7	37

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145	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
146	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
147	Reply: Evaluation of exome sequencing variation in undiagnosed ataxias. Brain, 2015, 138, e384-e384.	3.7	2
148	Use of stereotypical mutational motifs to define resolution limits for the ultra-deep resequencing of mitochondrial DNA. European Journal of Human Genetics, 2015, 23, 413-415.	1.4	10
149	Voxel-based analysis in neuroferritinopathy expands the phenotype and determines radiological correlates of disease severity. Journal of Neurology, 2015, 262, 2232-2240.	1.8	3
150	Clinical and Pathological Features of Mitochondrial DNA Deletion Disease Following Antiretroviral Treatment. JAMA Neurology, 2015, 72, 603.	4.5	3
151	SPG7 mutations are a common cause of undiagnosed ataxia. Neurology, 2015, 84, 1174-1176.	1.5	87
152	Phenotypic variability of TRPV4 related neuropathies. Neuromuscular Disorders, 2015, 25, 516-521.	0.3	27
153	Whole exome sequencing and the clinician: we need clinical skills and functional validation in variant filtering. Journal of Neurology, 2015, 262, 1673-1677.	1.8	14
154	Modifying Mitochondrial tRNAs: Delivering What the Cell Needs. Cell Metabolism, 2015, 21, 351-352.	7.2	6
155	Clinical heterogeneity of primary familial brain calcification due to a novel mutation in PDGFB. Neurology, 2015, 84, 1818-1820.	1.5	14
156	Retrospective assessment of the most common mitochondrial DNA mutations in a large Hungarian cohort of suspect mitochondrial cases. Mitochondrial DNA, 2015, 26, 572-578.	0.6	4
157	The p.Ser107Leu inBICD2is a mutation â€˜hot spotâ€™ causing distal spinal muscular atrophy. Brain, 2015, 138, e391-e391.	3.7	13
158	SCP2 mutations and neurodegeneration with brain iron accumulation. Neurology, 2015, 85, 1909-1911.	1.5	39
159	Electrophysiologic features of SYT2 mutations causing a treatable neuromuscular syndrome. Neurology, 2015, 85, 1964-1971.	1.5	47
160	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
161	Exome sequencing in undiagnosed inherited and sporadic ataxias. Brain, 2015, 138, 276-283.	3.7	120
162	Prevalence of neurogenetic disorders in the North of England. Neurology, 2015, 85, 1195-1201.	1.5	26

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163	Early-onset leukoencephalopathy due to a homozygous missense mutation in the DARS2 gene. Molecular and Cellular Probes, 2015, 29, 319-322.	0.9	14
164	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
165	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
166	ANO10 mutations cause ataxia and coenzyme Q10 deficiency. Journal of Neurology, 2014, 261, 2192-2198.	1.8	74
167	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
168	Treatable childhood neuronopathy caused by mutations in riboflavin transporter RFVT2. Brain, 2014, 137, 44-56.	3.7	143
169	Synaptotagmin 2 Mutations Cause an Autosomal-Dominant Form of Lambert-Eaton Myasthenic Syndrome and Nonprogressive Motor Neuropathy. American Journal of Human Genetics, 2014, 95, 472.	2.6	2
170	Founder p.Arg 446* mutation in the PDHX gene explains over half of cases with congenital lactic acidosis in Roma children. Molecular Genetics and Metabolism, 2014, 113, 76-83.	0.5	19
171	Riboflavin and CoQ Disorders. , 2014, , 233-244.		0
172	Chronic and slowly progressive weakness of the legs and hands. BMJ, The, 2014, 348, g459-g459.	3.0	6
173	EXOSC8 mutations alter mRNA metabolism and cause hypomyelination with spinal muscular atrophy and cerebellar hypoplasia. Nature Communications, 2014, 5, 4287.	5.8	120
174	Reply: Hereditary myopathy with early respiratory failure is caused by mutations in the titin FN3 119 domain. Brain, 2014, 137, e271-e271.	3.7	9
175	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
176	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
177	An under-recognised cause of spastic paraparesis in middle-aged women. Practical Neurology, 2014, 14, 182-184.	0.5	3
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