

Henna Riikka Susanna Tyynismaa

List of Publications by Year in descending order

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81
papers

4,864
citations

126907

33
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98798

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88
all docs

88
docs citations

88
times ranked

7257
citing authors

#	ARTICLE	IF	CITATIONS
1	FGF-21 as a biomarker for muscle-manifesting mitochondrial respiratory chain deficiencies: a diagnostic study. <i>Lancet Neurology</i> , The, 2011, 10, 806-818.	10.2	352
2	Mutant mitochondrial helicase Twinkle causes multiple mtDNA deletions and a late-onset mitochondrial disease in mice. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2005, 102, 17687-17692.	7.1	297
3	Mitochondrial myopathy induces a starvation-like response. <i>Human Molecular Genetics</i> , 2010, 19, 3948-3958.	2.9	249
4	Comparison of solution-based exome capture methods for next generation sequencing. <i>Genome Biology</i> , 2011, 12, R94.	9.6	237
5	Exome Sequencing Identifies Mitochondrial Alanine-tRNA Synthetase Mutations in Infantile Mitochondrial Cardiomyopathy. <i>American Journal of Human Genetics</i> , 2011, 88, 635-642.	6.2	229
6	Mitochondrial DNA Replication Defects Disturb Cellular dNTP Pools and Remodel One-Carbon Metabolism. <i>Cell Metabolism</i> , 2016, 23, 635-648.	16.2	222
7	Somatic Progenitor Cell Vulnerability to Mitochondrial DNA Mutagenesis Underlies Progeroid Phenotypes in Polg Mutator Mice. <i>Cell Metabolism</i> , 2012, 15, 100-109.	16.2	213
8	Twinkle helicase is essential for mtDNA maintenance and regulates mtDNA copy number. <i>Human Molecular Genetics</i> , 2004, 13, 3219-3227.	2.9	202
9	Ketogenic diet slows down mitochondrial myopathy progression in mice. <i>Human Molecular Genetics</i> , 2010, 19, 1974-1984.	2.9	168
10	Mitochondrial phenylalanyl-tRNA synthetase mutations underlie fatal infantile Alpers encephalopathy. <i>Human Molecular Genetics</i> , 2012, 21, 4521-4529.	2.9	143
11	Mitochondrial aminoacyl-tRNA synthetases in human disease. <i>Molecular Genetics and Metabolism</i> , 2013, 108, 206-211.	1.1	133
12	High mitochondrial DNA copy number has detrimental effects in mice. <i>Human Molecular Genetics</i> , 2010, 19, 2695-2705.	2.9	123
13	Twinkle mutations associated with autosomal dominant progressive external ophthalmoplegia lead to impaired helicase function and in vivo mtDNA replication stalling. <i>Human Molecular Genetics</i> , 2009, 18, 328-340.	2.9	120
14	De Novo Mutations in the Motor Domain of KIF1A Cause Cognitive Impairment, Spastic Paraparesis, Axonal Neuropathy, and Cerebellar Atrophy. <i>Human Mutation</i> , 2015, 36, 69-78.	2.5	114
15	A Heterozygous Truncating Mutation in RRM2B Causes Autosomal-Dominant Progressive External Ophthalmoplegia with Multiple mtDNA Deletions. <i>American Journal of Human Genetics</i> , 2009, 85, 290-295.	6.2	111
16	Human Heart Mitochondrial DNA Is Organized in Complex Catenated Networks Containing Abundant Four-way Junctions and Replication Forks. <i>Journal of Biological Chemistry</i> , 2009, 284, 21446-21457.	3.4	110
17	Overexpression of TFAM or Twinkle Increases mtDNA Copy Number and Facilitates Cardioprotection Associated with Limited Mitochondrial Oxidative Stress. <i>PLoS ONE</i> , 2015, 10, e0119687.	2.5	109
18	A locus for autosomal dominant keratoconus: linkage to 16q22.3-q23.1 in Finnish families. <i>Investigative Ophthalmology and Visual Science</i> , 2002, 43, 3160-4.	3.3	103

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19	Thymidine kinase 2 mutations in autosomal recessive progressive external ophthalmoplegia with multiple mitochondrial DNA deletions. <i>Human Molecular Genetics</i> , 2012, 21, 66-75.	2.9	91
20	Whole-exome sequencing identifies a mutation in the mitochondrial ribosome protein MRPL44 to underlie mitochondrial infantile cardiomyopathy. <i>Journal of Medical Genetics</i> , 2013, 50, 151-159.	3.2	85
21	Mouse models of mitochondrial DNA defects and their relevance for human disease. <i>EMBO Reports</i> , 2009, 10, 137-143.	4.5	84
22	Deficiency of the E3 ubiquitin ligase TRIM2 in early-onset axonal neuropathy. <i>Human Molecular Genetics</i> , 2013, 22, 2975-2983.	2.9	70
23	ATPase-deficient mitochondrial inner membrane protein ATAD3A disturbs mitochondrial dynamics in dominant hereditary spastic paraplegia. <i>Human Molecular Genetics</i> , 2017, 26, 1432-1443.	2.9	63
24	<i>CHCHD10</i> variant p.(Gly66Val) causes axonal Charcot-Marie-Tooth disease. <i>Neurology: Genetics</i> , 2015, 1, e1.	1.9	62
25	Liver Fat But Not Other Adiposity Measures Influence Circulating FGF21 Levels in Healthy Young Adult Twins. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2011, 96, E351-E355.	3.6	53
26	Serum angiopoietin-like 4 protein levels and expression in adipose tissue are inversely correlated with obesity in monozygotic twins. <i>Journal of Lipid Research</i> , 2011, 52, 1575-1582.	4.2	52
27	Selenoprotein biosynthesis defect causes progressive encephalopathy with elevated lactate. <i>Neurology</i> , 2015, 85, 306-315.	1.1	52
28	Loss of MICOS complex integrity and mitochondrial damage, but not TDP-43 mitochondrial localisation, are likely associated with severity of CHCHD10-related diseases. <i>Neurobiology of Disease</i> , 2018, 119, 159-171.	4.4	48
29	Structural modeling of tissue-specific mitochondrial alanyl-tRNA synthetase (AARS2) defects predicts differential effects on aminoacylation. <i>Frontiers in Genetics</i> , 2015, 6, 21.	2.3	46
30	Metabolic determination of cell fate through selective inheritance of mitochondria. <i>Nature Cell Biology</i> , 2022, 24, 148-154.	10.3	46
31	Dominant transmission of de novo KIF1A motor domain variant underlying pure spastic paraplegia. <i>European Journal of Human Genetics</i> , 2015, 23, 1427-1430.	2.8	44
32	Characterization of two unusual RS1 gene deletions segregating in Danish retinoschisis families. <i>Human Mutation</i> , 2000, 16, 307-314.	2.5	43
33	Enhanced cGAS-STING-dependent interferon signaling associated with mutations in ATAD3A. <i>Journal of Experimental Medicine</i> , 2021, 218, .	8.5	43
34	Identification of fifteen novel PHEX gene mutations in Finnish patients with hypophosphatemic rickets. <i>Human Mutation</i> , 2000, 15, 383-384.	2.5	39
35	Targeted next-generation sequencing reveals further genetic heterogeneity in axonal Charcot-Marie-Tooth neuropathy and a mutation in HSPB1. <i>European Journal of Human Genetics</i> , 2014, 22, 522-527.	2.8	33
36	Mitochondrial EFTs defects in juvenile-onset Leigh disease, ataxia, neuropathy, and optic atrophy. <i>Neurology</i> , 2014, 83, 743-751.	1.1	31

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37	MCM3AP in recessive Charcot-Marie-Tooth neuropathy and mild intellectual disability. <i>Brain</i> , 2017, 140, 2093-2103.	7.6	31
38	Editing activity for eliminating mischarged tRNAs is essential in mammalian mitochondria. <i>Nucleic Acids Research</i> , 2018, 46, 849-860.	14.5	30
39	Ribonucleotide reductase is not limiting for mitochondrial DNA copy number in mice. <i>Nucleic Acids Research</i> , 2010, 38, 8208-8218.	14.5	28
40	Dominant GDAP1 founder mutation is a common cause of axonal Charcot-Marie-Tooth disease in Finland. <i>Neurogenetics</i> , 2013, 14, 123-132.	1.4	28
41	Clinical and metabolic consequences of L-serine supplementation in hereditary sensory and autonomic neuropathy type 1C. <i>Journal of Physical Education and Sports Management</i> , 2017, 3, a002212.	1.2	27
42	Truncated HSPB1 causes axonal neuropathy and impairs tolerance to unfolded protein stress. <i>BBA Clinical</i> , 2015, 3, 233-242.	4.1	26
43	Capsid-Modified Adenoviral Vectors for Improved Muscle-Directed Gene Therapy. <i>Human Gene Therapy</i> , 2012, 23, 1065-1070.	2.7	25
44	Redox regulation of GRPEL2 nucleotide exchange factor for mitochondrial HSP70 chaperone. <i>Redox Biology</i> , 2018, 19, 37-45.	9.0	25
45	Absence of NEFL in patient-specific neurons in early-onset Charcot-Marie-Tooth neuropathy. <i>Neurology: Genetics</i> , 2018, 4, e244.	1.9	25
46	ALS and Parkinson's disease genes CHCHD10 and CHCHD2 modify synaptic transcriptomes in human iPSC-derived motor neurons. <i>Neurobiology of Disease</i> , 2020, 141, 104940.	4.4	24
47	Genetic background of HSH in three Polish families and a patient with an X;9 translocation. <i>European Journal of Human Genetics</i> , 2006, 14, 55-62.	2.8	23
48	Splicing Defect in Mitochondrial Seryl-tRNA Synthetase Gene Causes Progressive Spastic Paresis Instead of HUPRA Syndrome. <i>Human Mutation</i> , 2016, 37, 884-888.	2.5	23
49	TRIM2, a novel member of the antiviral family, limits New World arenavirus entry. <i>PLoS Biology</i> , 2019, 17, e3000137.	5.6	23
50	Neurofilament Light Regulates Axon Caliber, Synaptic Activity, and Organelle Trafficking in Cultured Human Motor Neurons. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 820105.	3.7	23
51	Clinical, biochemical, and genetic features associated with <i>VARs2</i> -related mitochondrial disease. <i>Human Mutation</i> , 2018, 39, 563-578.	2.5	22
52	<i>CHCHD10</i> mutations and motor neuron disease: the distribution in Finnish patients. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2017, 88, 272-277.	1.9	19
53	Instability of the mitochondrial alanyl-tRNA synthetase underlies fatal infantile-onset cardiomyopathy. <i>Human Molecular Genetics</i> , 2019, 28, 258-268.	2.9	19
54	The Variant p.(Arg183Trp) in SPTLC2 Causes Late-Onset Hereditary Sensory Neuropathy. <i>NeuroMolecular Medicine</i> , 2016, 18, 81-90.	3.4	18

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55	The Overexpression of Twinkle Helicase Ameliorates the Progression of Cardiac Fibrosis and Heart Failure in Pressure Overload Model in Mice. <i>PLoS ONE</i> , 2013, 8, e67642.	2.5	18
56	Exposure to arginine analog canavanine induces aberrant mitochondrial translation products, mitoribosome stalling, and instability of the mitochondrial proteome. <i>International Journal of Biochemistry and Cell Biology</i> , 2015, 65, 268-274.	2.8	16
57	Effectiveness of clinical exome sequencing in adult patients with difficult-to-diagnose neurological disorders. <i>Acta Neurologica Scandinavica</i> , 2022, 145, 63-72.	2.1	16
58	Recessive PYROXD1 mutations cause adult-onset limb-girdle-type muscular dystrophy. <i>Journal of Neurology</i> , 2019, 266, 353-360.	3.6	15
59	Mouse models of mtDNA replication diseases. <i>Methods</i> , 2010, 51, 405-410.	3.8	12
60	Mixing and matching mitochondrial aminoacyl synthetases and their tRNA s: a new way to treat respiratory chain disorders?. <i>EMBO Molecular Medicine</i> , 2014, 6, 155-157.	6.9	12
61	Screening for Fabry disease and Hereditary ATTR amyloidosis in idiopathic small fiber and mixed neuropathy. <i>Muscle and Nerve</i> , 2019, 59, 354-357.	2.2	12
62	PFKM gene defect and glycogen storage disease GSDVII with misleading enzyme histochemistry. <i>Neurology: Genetics</i> , 2015, 1, e7.	1.9	11
63	A patient with pontocerebellar hypoplasia type 6: Novel RARS2 mutations, comparison to previously published patients and clinical distinction from PEHO syndrome. <i>European Journal of Medical Genetics</i> , 2020, 63, 103766.	1.3	10
64	Unique Exercise Lactate Profile in Muscle Phosphofructokinase Deficiency (Tarui Disease); Difference Compared with McArdle Disease. <i>Frontiers in Neurology</i> , 2016, 7, 82.	2.4	9
65	Dominant mutations in ITPR3 cause Charcot-Marie-Tooth disease. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 1962-1972.	3.7	9
66	Threshold of heteroplasmic truncating MT-ATP6 mutation in reprogramming, Notch hyperactivation and motor neuron metabolism. <i>Human Molecular Genetics</i> , 2022, 31, 958-974.	2.9	9
67	<i>De novo</i> SPTAN1 mutation in axonal sensorimotor neuropathy and developmental disorder. <i>Brain</i> , 2020, 143, e104-e104.	7.6	8
68	Bi-allelic loss-of-function OBSCN variants predispose individuals to severe recurrent rhabdomyolysis. <i>Brain</i> , 2022, 145, 3985-3998.	7.6	6
69	Structural insights into Charcot-Marie-Tooth disease-linked mutations in human GDAP1. <i>FEBS Open Bio</i> , 2022, 12, 1306-1324.	2.3	6
70	Severe neonatal MEGDH syndrome with a homozygous truncating mutation in SERAC1. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2022, 1868, 166298.	3.8	5
71	Attitudes towards genetic testing and information: does parenthood shape the views?. <i>Journal of Community Genetics</i> , 2020, 11, 461-473.	1.2	4
72	Distinct effects on mRNA export factor GANP underlie neurological disease phenotypes and alter gene expression depending on intron content. <i>Human Molecular Genetics</i> , 2020, 29, 1426-1439.	2.9	4

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73	Mitochondrial Aminoacyl-tRNA Synthetases. , 2013, , 263-276.		4
74	Serum Creatine, Not Neurofilament Light, Is Elevated in CHCHD10-Linked Spinal Muscular Atrophy. Frontiers in Neurology, 2022, 13, 793937.	2.4	4
75	Comparative whole-genome transcriptome analysis in renal cell populations reveals high tissue specificity of MAPK/ERK targets in embryonic kidney. BMC Biology, 2022, 20, 112.	3.8	4
76	Reply: A novel MCM3AP mutation in a Lebanese family with recessive Charcot-Marie-Tooth neuropathy. Brain, 2018, 141, e67-e67.	7.6	3
77	Generating Mouse Models of Mitochondrial Disease. , 2015, , 689-701.		2
78	Analysis of Mitochondrial Protein Synthesis: De Novo Translation, Steady-State Levels, and Assembled OXPHOS Complexes. Current Protocols in Toxicology / Editorial Board, Mahin D Maines (editor-in-chief) [et Al], 2018, 77, e56.	1.1	2
79	A Metabolic Vulnerability of Vision. New England Journal of Medicine, 2019, 381, 1474-1476.	27.0	2
80	LATE BREAKING NEWS E-POSTER PRESENTATION. Neuromuscular Disorders, 2020, 30, S167-S168.	0.6	0
81	The Increase of Mitochondrial DNA Copy Number Attenuates Eccentric Cardiac Remodeling In Volume Overload Model. FASEB Journal, 2013, 27, 1129.11.	0.5	0