Henna Riikka Susanna Tyynismaa

List of Publications by Year
in descending order

81 papers	4,864 citations	126907 33 h-index	98798 67 g-index
88 all docs	88 docs citations	88 times ranked	7257 citing authors

#	Article	IF	Citations
1	Effectiveness of clinical exome sequencing in adult patients with difficultâ€toâ€diagnose neurological disorders. Acta Neurologica Scandinavica, 2022, 145, 63-72.	2.1	16
2	Threshold of heteroplasmic truncating MT-ATP6 mutation in reprogramming, Notch hyperactivation and motor neuron metabolism. Human Molecular Genetics, 2022, 31, 958-974.	2.9	9
3	Severe neonatal MEGDHEL syndrome with a homozygous truncating mutation in SERAC1. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2022, 1868, 166298.	3.8	5
4	Serum Creatine, Not Neurofilament Light, Is Elevated in CHCHD10-Linked Spinal Muscular Atrophy. Frontiers in Neurology, 2022, 13, 793937.	2.4	4
5	Metabolic determination of cell fate through selective inheritance of mitochondria. Nature Cell Biology, 2022, 24, 148-154.	10.3	46
6	Bi-allelic loss-of-function OBSCN variants predispose individuals to severe recurrent rhabdomyolysis. Brain, 2022, 145, 3985-3998.	7.6	6
7	Structural insights into Charcotâ€“Marieâ€“Tooth diseaseâ€linked mutations in human GDAP1. FEBS Open Bio, 2022, 12, 1306-1324.	2.3	6
8	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
9	Enhanced cGAS-STINGâ€“dependent interferon signaling associated with mutations in ATAD3A. Journal of Experimental Medicine, 2021, 218, .	8.5	43
10	Neurofilament Light Regulates Axon Caliber, Synaptic Activity, and Organelle Trafficking in Cultured Human Motor Neurons. Frontiers in Cell and Developmental Biology, 2021, 9, 820105.	3.7	23
11	A patient with pontocerebellar hypoplasia type 6: Novel RARS2 mutations, comparison to previously published patients and clinical distinction from PEHO syndrome. European Journal of Medical Genetics, 2020, 63, 103766.	1.3	10
12	LATE BREAKING NEWS E-POSTER PRESENTATION. Neuromuscular Disorders, 2020, 30, S167-S168.	0.6	0
13	De novo SPTAN1mutation in axonal sensorimotor neuropathy and developmental disorder. Brain, 2020, 143, e104-e104.	7.6	8
14	Dominant mutations in ITPR3 cause Charcotâ€Marieâ€Tooth disease. Annals of Clinical and Translational Neurology, 2020, 7, 1962-1972.	3.7	9
15	Attitudes towards genetic testing and information: does parenthood shape the views?. Journal of Community Genetics, 2020, 11, 461-473.	1.2	4
16	Distinct effects on mRNA export factor GANP underlie neurological disease phenotypes and alter gene expression depending on intron content. Human Molecular Genetics, 2020, 29, 1426-1439.	2.9	4
17	ALS and Parkinson's disease genes CHCHD10 and CHCHD2 modify synaptic transcriptomes in human iPSC-derived motor neurons. Neurobiology of Disease, 2020, 141, 104940.	4.4	24
18	Instability of the mitochondrial alanyl-tRNA synthetase underlies fatal infantile-onset cardiomyopathy. Human Molecular Genetics, 2019, 28, 258-268.	2.9	19

#	Article	IF	Citations
19	A Metabolic Vulnerability of Vision. New England Journal of Medicine, 2019, 381, 1474-1476.	27.0	2
20	TRIM2, a novel member of the antiviral family, limits New World arenavirus entry. PLoS Biology, 2019, 17, e3000137.	5.6	23
21	Recessive PYROXD1 mutations cause adult-onset limb-girdle-type muscular dystrophy. Journal of Neurology, 2019, 266, 353-360.	3.6	15
22	Screening for Fabry disease and Hereditary ATTR amyloidosis in idiopathic smallâ€fiber and mixed neuropathy. Muscle and Nerve, 2019, 59, 354-357.	2.2	12
23	Clinical, biochemical, and genetic features associated with VARS2 -related mitochondrial disease. Human Mutation, 2018, 39, 563-578.	2.5	22
24	Editing activity for eliminating mischarged tRNAs is essential in mammalian mitochondria. Nucleic Acids Research, 2018, 46, 849-860.	14.5	30
25	Reply: A novel MCM3AP mutation in a Lebanese family with recessive Charcot-Marie-Tooth neuropathy. Brain, 2018, 141, e67-e67.	7.6	3
26	Analysis of Mitochondrial Protein Synthesis: De Novo Translation, Steadyâ€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
27	Redox regulation of GRPEL2 nucleotide exchange factor for mitochondrial HSP70 chaperone. Redox Biology, 2018, 19, 37-45.	9.0	25
28	Loss of MICOS complex integrity and mitochondrial damage, but not TDP-43 mitochondrial localisation, are likely associated with severity of CHCHD10-related diseases. Neurobiology of Disease, 2018, 119, 159-171.	4.4	48
29	Absence of NEFL in patient-specific neurons in early-onset Charcot-Marie-Tooth neuropathy. Neurology: Genetics, 2018, 4, e244.	1.9	25
30	CHCHD10 mutations and motor neuron disease: the distribution in Finnish patients. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 272-277.	1.9	19
31	MCM3AP in recessive Charcot-Marie-Tooth neuropathy and mild intellectual disability. Brain, 2017, 140, 2093-2103.	7.6	31
32	Clinical and metabolic consequences of L-serine supplementation in hereditary sensory and autonomic neuropathy type 1C. Journal of Physical Education and Sports Management, 2017, 3, a002212.	1.2	27
33	ATPase-deficient mitochondrial inner membrane protein ATAD3A disturbs mitochondrial dynamics in dominant hereditary spastic paraplegia. Human Molecular Genetics, 2017, 26, 1432-1443.	2.9	63
34	Unique Exercise Lactate Profile in Muscle Phosphofructokinase Deficiency (Tarui Disease); Difference Compared with McArdle Disease. Frontiers in Neurology, 2016, 7, 82.	2.4	9
35	Splicing Defect in Mitochondrial Seryl-tRNA Synthetase Gene Causes Progressive Spastic Paresis Instead of HUPRA Syndrome. Human Mutation, 2016, 37, 884-888.	2.5	23
36	Mitochondrial DNA Replication Defects Disturb Cellular dNTP Pools and Remodel One-Carbon Metabolism. Cell Metabolism, 2016, 23, 635-648.	16.2	222

#	Article	IF	Citations
37	The Variant p.(Arg183Trp) in SPTLC2 Causes Late-Onset Hereditary Sensory Neuropathy. NeuroMolecular Medicine, 2016, 18, 81-90.	3.4	18
38	CHCHD10 variant p.(Gly66Val) causes axonal Charcot-Marie-Tooth disease. Neurology: Genetics, 2015, 1, e1.	1.9	62
39	Overexpression of TFAM or Twinkle Increases mtDNA Copy Number and Facilitates Cardioprotection Associated with Limited Mitochondrial Oxidative Stress. PLoS ONE, 2015, 10, e0119687.	2.5	109
40	De Novo Mutations in the Motor Domain of KIF1A Cause Cognitive Impairment, Spastic Paraparesis, Axonal Neuropathy, and Cerebellar Atrophy. Human Mutation, 2015, 36, 69-78.	2.5	114
41	Dominant transmission of de novo KIF1A motor domain variant underlying pure spastic paraplegia. European Journal of Human Genetics, 2015, 23, 1427-1430.	2.8	44
42	Structural modeling of tissue-specific mitochondrial alanyl-tRNA synthetase (AARS2) defects predicts differential effects on aminoacylation. Frontiers in Genetics, 2015, 6, 21.	2.3	46
43	Selenoprotein biosynthesis defect causes progressive encephalopathy with elevated lactate. Neurology, 2015, 85, 306-315.	1.1	52
44	Exposure to arginine analog canavanine induces aberrant mitochondrial translation products, mitoribosome stalling, and instability of the mitochondrial proteome. International Journal of Biochemistry and Cell Biology, 2015, 65, 268-274.	2.8	16
45	Generating Mouse Models of Mitochondrial Disease. , 2015, , 689-701.		2
46	Truncated HSPB1 causes axonal neuropathy and impairs tolerance to unfolded protein stress. BBA Clinical, 2015, 3, 233-242.	4.1	26
47	PFKMgene defect and glycogen storage disease GSDVII with misleading enzyme histochemistry. Neurology: Genetics, 2015, 1, e7.	1.9	11
48	Mitochondrial EFTs defects in juvenile-onset Leigh disease, ataxia, neuropathy, and optic atrophy. Neurology, 2014, 83, 743-751.	1.1	31
49	Mixing and matching mitochondrial aminoacyl synthetases and their tRNA s: a new way to treat respiratory chain disorders?. EMBO Molecular Medicine, 2014, 6, 155-157.	6.9	12
50	Targeted next-generation sequencing reveals further genetic heterogeneity in axonal Charcotâ€“Marieâ€“Tooth neuropathy and a mutation in HSPB1. European Journal of Human Genetics, 2014, 22, 522-527.	2.8	33
51	Dominant GDAP1 founder mutation is a common cause of axonal Charcot-Marie-Tooth disease in Finland. Neurogenetics, 2013, 14, 123-132.	1.4	28
52	Mitochondrial aminoacyl-tRNA synthetases in human disease. Molecular Genetics and Metabolism, 2013, 108, 206-211.	1.1	133
53	Deficiency of the E3 ubiquitin ligase TRIM2 in early-onset axonal neuropathy. Human Molecular Genetics, 2013, 22, 2975-2983.	2.9	70
54	Whole-exome sequencing identifies a mutation in the mitochondrial ribosome protein MRPL44 to underlie mitochondrial infantile cardiomyopathy. Journal of Medical Genetics, 2013, 50, 151-159.	3.2	85

#	Article	IF	Citations
55	Mitochondrial Aminoacyl-tRNA Synthetases. , 2013, , 263-276.		4
56	The Overexpression of Twinkle Helicase Ameliorates the Progression of Cardiac Fibrosis and Heart Failure in Pressure Overload Model in Mice. PLoS ONE, 2013, 8, e67642.	2.5	18
57	The Increase of Mitochondrial DNA Copy Number Attenuates Eccentric Cardiac Remodeling In Volume Overload Model. FASEB Journal, 2013, 27, 1129.11.	0.5	0
58	Thymidine kinase 2 mutations in autosomal recessive progressive external ophthalmoplegia with multiple mitochondrial DNA deletions. Human Molecular Genetics, 2012, 21, 66-75.	2.9	91
59	Capsid-Modified Adenoviral Vectors for Improved Muscle-Directed Gene Therapy. Human Gene Therapy, 2012, 23, 1065-1070.	2.7	25
60	Mitochondrial phenylalanyl-tRNA synthetase mutations underlie fatal infantile Alpers encephalopathy. Human Molecular Genetics, 2012, 21, 4521-4529.	2.9	143
61	Somatic Progenitor Cell Vulnerability to Mitochondrial DNA Mutagenesis Underlies Progeroid Phenotypes in Polg Mutator Mice. Cell Metabolism, 2012, 15, 100-109.	16.2	213
62	Comparison of solution-based exome capture methods for next generation sequencing. Genome Biology, 2011, 12, R94.	9.6	237
63	FGF-21 as a biomarker for muscle-manifesting mitochondrial respiratory chain deficiencies: a diagnostic study. Lancet Neurology, The, 2011, 10, 806-818.	10.2	352
64	Exome Sequencing Identifies Mitochondrial Alanyl-tRNA Synthetase Mutations in Infantile Mitochondrial Cardiomyopathy. American Journal of Human Genetics, 2011, 88, 635-642.	6.2	229
65	Liver Fat But Not Other Adiposity Measures Influence Circulating FGF21 Levels in Healthy Young Adult Twins. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E351-E355.	3.6	53
66	Serum angiopoietin-like 4 protein levels and expression in adipose tissue are inversely correlated with obesity in monozygotic twins. Journal of Lipid Research, 2011, 52, 1575-1582.	4.2	52
67	Ribonucleotide reductase is not limiting for mitochondrial DNA copy number in mice. Nucleic Acids Research, 2010, 38, 8208-8218.	14.5	28
68	Ketogenic diet slows down mitochondrial myopathy progression in mice. Human Molecular Genetics, 2010, 19, 1974-1984.	2.9	168
69	High mitochondrial DNA copy number has detrimental effects in mice. Human Molecular Genetics, 2010, 19, 2695-2705.	2.9	123
70	Mitochondrial myopathy induces a starvation-like response. Human Molecular Genetics, 2010, 19, 3948-3958.	2.9	249
71	Mouse models of mtDNA replication diseases. Methods, 2010, 51, 405-410.	3.8	12
72	Human Heart Mitochondrial DNA Is Organized in Complex Catenated Networks Containing Abundant Four-way Junctions and Replication Forks. Journal of Biological Chemistry, 2009, 284, 21446-21457.	3.4	110

#	Article	IF	Citations
73	Twinkle mutations associated with autosomal dominant progressive external ophthalmoplegia lead to impaired helicase function and in vivo mtDNA replication stalling. Human Molecular Genetics, 2009, 18, 328-340.	2.9	120
74	Mouse models of mitochondrial DNA defects and their relevance for human disease. EMBO Reports, 2009, 10, 137-143.	4.5	84
75	A Heterozygous Truncating Mutation in RRM2B Causes Autosomal-Dominant Progressive External Ophthalmoplegia with Multiple mtDNA Deletions. American Journal of Human Genetics, 2009, 85, 290-295.	6.2	111
76	Genetic background of HSH in three Polish families and a patient with an X;9 translocation. European Journal of Human Genetics, 2006, 14, 55-62.	2.8	23
77	Mutant mitochondrial helicase Twinkle causes multiple mtDNA deletions and a late-onset mitochondrial disease in mice. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 17687-17692.	7.1	297
78	Twinkle helicase is essential for mtDNA maintenance and regulates mtDNA copy number. Human Molecular Genetics, 2004, 13, 3219-3227.	2.9	202
79	A locus for autosomal dominant keratoconus: linkage to 16q22.3-q23.1 in Finnish families. Investigative Ophthalmology and Visual Science, 2002, 43, 3160-4.	3.3	103
80	Identification of fifteen novel PHEX gene mutations in Finnish patients with hypophosphatemic rickets. Human Mutation, 2000, 15, 383-384.	2.5	39
81	Characterization of two unusual RS1 gene deletions segregating in Danish retinoschisis families. Human Mutation, 2000, 16, 307-314.	2.5	43