Anu Suomalainen

List of Publications by Year
in descending order

127	15,570	25034 57	18647 119
papers	citations	h-index	g-index

135	135	135	17100
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mitochondria: In Sickness and in Health. Cell, 2012, 148, 1145-1159.	28.9	2,411
2	Mitochondrial diseases. Nature Reviews Disease Primers, 2016, 2, 16080.	30.5	1,001
3	Human mitochondrial DNA deletions associated with mutations in the gene encoding Twinkle, a phage T7 gene 4-like protein localized in mitochondria. Nature Genetics, 2001, 28, 223-231.	21.4	803
4	Role of Adenine Nucleotide Translocator 1 in mtDNA Maintenance. Science, 2000, 289, 782-785.	12.6	591
5	Parkinsonism, premature menopause, and mitochondrial DNA polymerase Î³ mutations: clinical and molecular genetic study. Lancet, The, 2004, 364, 875-882.	13.7	538
6	Phenotypic spectrum associated with mutations of the mitochondrial polymerase Â gene. Brain, 2006, 129, 1674-1684.	7.6	397
7	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.	7.6	381
8	Mitochondrial diseases: the contribution of organelle stress responses to pathology. Nature Reviews Molecular Cell Biology, 2018, 19, 77-92.	37.0	369
9	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
10	Effective treatment of mitochondrial myopathy by nicotinamide riboside, a vitamin <scp>B</scp>3. EMBO Molecular Medicine, 2014, 6, 721-731.	6.9	326
11	Mitochondrial DNA Polymerase W748S Mutation: A Common Cause of Autosomal Recessive Ataxia with Ancient European Origin. American Journal of Human Genetics, 2005, 77, 430-441.	6.2	302
12	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
13	mTORC1 Regulates Mitochondrial Integrated Stress Response and Mitochondrial Myopathy Progression. Cell Metabolism, 2017, 26, 419-428.e5.	16.2	291
14	GRACILE Syndrome, a Lethal Metabolic Disorder with Iron Overload, Is Caused by a Point Mutation in BCS1L. American Journal of Human Genetics, 2002, 71, 863-876.	6.2	263
15	Impaired Mitochondrial Biogenesis in Adipose Tissue in Acquired Obesity. Diabetes, 2015, 64, 3135-3145.	0.6	263
16	Mitochondrial myopathy induces a starvation-like response. Human Molecular Genetics, 2010, 19, 3948-3958.	2.9	249
17	Comparison of solution-based exome capture methods for next generation sequencing. Genome Biology, 2011, 12, R94.	9.6	237
18	Exome Sequencing Identifies Mitochondrial Alanyl-tRNA Synthetase Mutations in Infantile Mitochondrial Cardiomyopathy. American Journal of Human Genetics, 2011, 88, 635-642.	6.2	229

#	Article	IF	Citations
19	Mitochondrial DNA Replication Defects Disturb Cellular dNTP Pools and Remodel One-Carbon Metabolism. Cell Metabolism, 2016, 23, 635-648.	16.2	222
20	Somatic Progenitor Cell Vulnerability to Mitochondrial DNA Mutagenesis Underlies Progeroid Phenotypes in Polg Mutator Mice. Cell Metabolism, 2012, 15, 100-109.	16.2	213
21	Twinkle helicase is essential for mtDNA maintenance and regulates mtDNA copy number. Human Molecular Genetics, 2004, 13, 3219-3227.	2.9	202
22	Infantile onset spinocerebellar ataxia is caused by recessive mutations in mitochondrial proteins Twinkle and Twinky. Human Molecular Genetics, 2005, 14, 2981-2990.	2.9	201
23	Recessive Twinkle mutations in early onset encephalopathy with mtDNA depletion. Brain, 2007, 130, 3032-3040.	7.6	188
24	Tissue- and cell-typeâ€“specific manifestations of heteroplasmic mtDNA 3243A>G mutation in human induced pluripotent stem cell-derived disease model. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, E3622-30.	7.1	185
25	Mitochondrial DNA depletion syndromes â€“ Many genes, common mechanisms. Neuromuscular Disorders, 2010, 20, 429-437.	0.6	169
26	Ketogenic diet slows down mitochondrial myopathy progression in mice. Human Molecular Genetics, 2010, 19, 1974-1984.	2.9	168
27	FGF21 is a biomarker for mitochondrial translation and mtDNA maintenance disorders. Neurology, 2016, 87, 2290-2299.	1.1	167
28	Fibroblast Growth Factor 21 Drives Dynamics of Local and Systemic Stress Responses in Mitochondrial Myopathy with mtDNA Deletions. Cell Metabolism, 2019, 30, 1040-1054.e7.	16.2	166
29	Niacin Cures Systemic NAD+ Deficiency and Improves Muscle Performance in Adult-Onset Mitochondrial Myopathy. Cell Metabolism, 2020, 31, 1078-1090.e5.	16.2	154
30	Mechanisms of mitochondrial diseases. Annals of Medicine, 2012, 44, 41-59.	3.8	149
31	Infantile-onset spinocerebellar ataxia and mitochondrial recessive ataxia syndrome are associated with neuronal complex I defect and mtDNA depletion. Human Molecular Genetics, 2008, 17, 3822-3835.	2.9	129
32	Effect of bezafibrate treatment on late-onset mitochondrial myopathy in mice. Human Molecular Genetics, 2012, 21, 526-535.	2.9	125
33	High mitochondrial DNA copy number has detrimental effects in mice. Human Molecular Genetics, 2010, 19, 2695-2705.	2.9	123
34	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
35	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
36	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
37	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
38	Twinkle and POLG defects enhance age-dependent accumulation of mutations in the control region of mtDNA. Nucleic Acids Research, 2004, 32, 3053-3064.	14.5	107
39	Diseases caused by nuclear genes affecting mtDNA stability. American Journal of Medical Genetics Part A, 2001, 106, 53-61.	2.4	100
40	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.	6.2	99
41	Genetic Basis of Severe Childhood-OnsetÂCardiomyopathies. Journal of the American College of Cardiology, 2018, 72, 2324-2338.	2.8	97
42	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.	2.9	96
43	Mitochondrial disease in adults: recent advances and future promise. Lancet Neurology, The, 2021, 20, 573-584.	10.2	96
44	Recurrent De Novo Dominant Mutations in SLC25A4 Cause Severe Early-Onset Mitochondrial Disease and Loss of Mitochondrial DNA Copy Number. American Journal of Human Genetics, 2016, 99, 860-876.	6.2	93
45	Abundance of the POLG disease mutations in Europe, Australia, New Zealand, and the United States explained by single ancient European founders. European Journal of Human Genetics, 2007, 15, 779-783.	2.8	91
46	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
47	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
48	Mouse models of mitochondrial DNA defects and their relevance for human disease. EMBO Reports, 2009, 10, 137-143.	4.5	84
49	New mutation of mitochondrial DNAJC19 causing dilated and noncompaction cardiomyopathy, anemia, ataxia, and male genital anomalies. Pediatric Research, 2012, 72, 432-437.	2.3	83
50	Phosphorylation of Parkin at serine 65 is essential for its activationin vivo. Open Biology, 2018, 8, 180108.	3.6	81
51	TFPa/HADHA is required for fatty acid beta-oxidation and cardiolipin re-modeling in human cardiomyocytes. Nature Communications, 2019, 10, 4671.	12.8	77
52	Thymidine kinase 2 defects can cause multi-tissue mtDNA depletion syndrome. Brain, 2008, 131, 2841-2850.	7.6	73
53	Retrospective natural history of thymidine kinase 2 deficiency. Journal of Medical Genetics, 2018, 55, 515-521.	3.2	73
54	Quantification of tRNA3243Leu point mutation of mitochondrial DNA in MELAS patients and its effects on mitochondrial transcription. Human Molecular Genetics, 1993, 2, 525-534.	2.9	72

#	Article	IF	Citations
55	mtDNA Mutagenesis Disrupts Pluripotent Stem Cell Function by Altering Redox Signaling. Cell Reports, 2015, 11, 1614-1624.	6.4	66
56	Regulation of Mother-to-Offspring Transmission of mtDNA Heteroplasmy. Cell Metabolism, 2019, 30, 1120-1130.e5.	16.2	66
57	Stem cells, mitochondria and aging. Biochimica Et Biophysica Acta - Bioenergetics, 2015, 1847, 1380-1386.	1.0	65
58	RNA modification landscape of the human mitochondrial tRNALys regulates protein synthesis. Nature Communications, 2018, 9, 3966.	12.8	61
59	Deficiency of the INCL protein Ppt1 results in changes in ectopic F1-ATP synthase and altered cholesterol metabolism. Human Molecular Genetics, 2008, 17, 1406-1417.	2.9	58
60	USF1 deficiency activates brown adipose tissue and improves cardiometabolic health. Science Translational Medicine, 2016, 8, 323ra13.	12.4	58
61	Defects in mtDNA replication challenge nuclear genome stability through nucleotide depletion and provide a unifying mechanism for mouse progerias. Nature Metabolism, 2019, 1, 958-965.	11.9	57
62	Modified Atkins diet induces subacute selective raggedâ€redâ€fiber lysis in mitochondrial myopathyÂpatients. EMBO Molecular Medicine, 2016, 8, 1234-1247.	6.9	56
63	Metabolomes of mitochondrial diseases and inclusion body myositis patients: treatment targets and biomarkers. EMBO Molecular Medicine, 2018, 10, .	6.9	54
64	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
65	Selenoprotein biosynthesis defect causes progressive encephalopathy with elevated lactate. Neurology, 2015, 85, 306-315.	1.1	52
66	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
67	Fibroblast growth factor 21: a novel biomarker for human muscle-manifesting mitochondrial disorders. Expert Opinion on Medical Diagnostics, 2013, 7, 313-317.	1.6	45
68	Clustering of Alpers disease mutations and catalytic defects in biochemical variants reveal new features of molecular mechanism of the human mitochondrial replicase, Pol Î³. Nucleic Acids Research, 2011, 39, 9072-9084.	14.5	44
69	Vegan diet in young children remodels metabolism and challenges the statuses of essential nutrients. EMBO Molecular Medicine, 2021, 13, e13492.	6.9	43
70	Therapy for mitochondrial disorders: Little proof, high research activity, some promise. Seminars in Fetal and Neonatal Medicine, 2011, 16, 236-240.	2.3	41
71	Biomarkers for mitochondrial respiratory chain disorders. Journal of Inherited Metabolic Disease, 2011, 34, 277-282.	3.6	41
72	Mesencephalic complex I deficiency does not correlate with parkinsonism in mitochondrial DNA maintenance disorders. Brain, 2013, 136, 2379-2392.	7.6	41

#	Article	IF	Citations
73	Defective mitochondrial RNA processing due to PNPT1 variants causes Leigh syndrome. Human Molecular Genetics, 2017, 26, 3352-3361.	2.9	41
74	Overexpression of Twinkle-helicase protects cardiomyocytes from genotoxic stress caused by reactive oxygen species. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 19408-19413.	7.1	39
75	Integrative omics approaches provide biological and clinical insights: examples from mitochondrial diseases. Journal of Clinical Investigation, 2020, 130, 20-28.	8.2	39
76	Characterization of a novel human putative mitochondrial transporter homologous to the yeast mitochondrial RNA splicing proteins 3 and 4. FEBS Letters, 2001, 494, 79-84.	2.8	38
77	Loss of mtDNA activates astrocytes and leads to spongiotic encephalopathy. Nature Communications, 2018, 9, 70.	12.8	38
78	SUCLA2 mutations cause global protein succinylation contributing to the pathomechanism of a hereditary mitochondrial disease. Nature Communications, 2020, 11, 5927.	12.8	35
79	Mosaic dysfunction of mitophagy in mitochondrial muscle disease. Cell Metabolism, 2022, 34, 197-208.e5.	16.2	35
80	Diagnostic value of serum biomarkers <scp>FGF21</scp> and <scp>GDF15</scp> compared to muscle sample in mitochondrial disease. Journal of Inherited Metabolic Disease, 2021, 44, 469-480.	3.6	34
81	A variant inMRPS14(uS14m) causes perinatal hypertrophic cardiomyopathy with neonatal lactic acidosis, growth retardation, dysmorphic features and neurological involvement. Human Molecular Genetics, 2019, 28, 639-649.	2.9	33
82	Mitochondrial EFTs defects in juvenile-onset Leigh disease, ataxia, neuropathy, and optic atrophy. Neurology, 2014, 83, 743-751.	1.1	31
83	Quantitative Analysis of Human DNA Sequences by PCR and Solid-Phase Minisequencing. Molecular Biotechnology, 2000, 15, 123-132.	2.4	29
84	Patient-Specific Induced Pluripotent Stem Cellâ€“Derived RPE Cells: Understanding the Pathogenesis of Retinopathy in Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency. , 2015, 56, 3371.		29
85	Ribonucleotide reductase is not limiting for mitochondrial DNA copy number in mice. Nucleic Acids Research, 2010, 38, 8208-8218.	14.5	28
86	Mitochondrial stress response triggered by defects in protein synthesis quality control. Life Science Alliance, 2019, 2, e201800219.	2.8	26
87	Differential metabolic consequences of fumarate hydratase and respiratory chain defects. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2008, 1782, 287-294.	3.8	23
88	Mitochondrial encephalomyopathy and retinoblastoma explained by compound heterozygosity of SUCLA2 point mutation and 13q14 deletion. European Journal of Human Genetics, 2015, 23, 325-330.	2.8	20
89	Instability of the mitochondrial alanyl-tRNA synthetase underlies fatal infantile-onset cardiomyopathy. Human Molecular Genetics, 2019, 28, 258-268.	2.9	19
90	Mitochondrial recessive ataxia syndrome mimicking dominant spinocerebellar ataxia. Journal of the Neurological Sciences, 2012, 315, 160-163.	0.6	18

#	Article	IF	Citations
91	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
92	IMPDH2: a new gene associated with dominant juvenile-onset dystonia-tremor disorder. European Journal of Human Genetics, 2021, 29, 1833-1837.	2.8	17
93	A urinary biosignature for mitochondrial myopathy, encephalopathy, lactic acidosis and stroke like episodes (MELAS). Mitochondrion, 2019, 45, 38-45.	3.4	16
94	<scp>MED27</scp> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. Annals of Neurology, 2021, 89, 828-833.	5.3	14
95	Mouse models of mtDNA replication diseases. Methods, 2010, 51, 405-410.	3.8	12
96	Mitochondrial spongiotic brain disease: astrocytic stress and harmful rapamycin and ketosis effect. Life Science Alliance, 2020, 3, e202000797.	2.8	12
97	Cost-effectiveness of whole-exome sequencing in progressive neurological disorders of children. European Journal of Paediatric Neurology, 2022, 36, 30-36.	1.6	12
98	Mitochondrial roles in disease: a box full of surprises. EMBO Molecular Medicine, 2015, 7, 1245-1247.	6.9	11
99	Absence of Hikeshi, a nuclear transporter for heat-shock protein HSP70, causes infantile hypomyelinating leukoencephalopathy. European Journal of Human Genetics, 2017, 25, 366-370.	2.8	11
100	Using urine to diagnose largeâ€scale mtDNA deletions in adult patients. Annals of Clinical and Translational Neurology, 2020, 7, 1318-1326.	3.7	11
101	The rare Costello variant HRAS c.173C>T (p.T58I) with severe neonatal hypertrophic cardiomyopathy. American Journal of Medical Genetics, Part A, 2016, 170, 1433-1438.	1.2	10
102	In-frame deletion in canine PITRM1 is associated with a severe early-onset epilepsy, mitochondrial dysfunction and neurodegeneration. Human Genetics, 2021, 140, 1593-1609.	3.8	9
103	Asymmetric rejuvenation. Nature, 2015, 521, 296-298.	27.8	8
104	A complex genomic locus drives mt DNA replicase POLG expression to its diseaseâ€related nervous system regions. EMBO Molecular Medicine, 2018, 10, 13-21.	6.9	8
105	Disruption of the mouse Shmt2 gene confers embryonic anaemia via foetal liver-specific metabolomic disorders. Scientific Reports, 2019, 9, 16054.	3.3	8
106	Phenotypic effects of dietary stress in combination with a respiratory chain bypass in mice. Physiological Reports, 2019, 7, e14159.	1.7	8
107	The relevance of mitochondrial DNA variants fluctuation during reprogramming and neuronal differentiation of human iPSCs. Stem Cell Reports, 2021, 16, 1953-1967.	4.8	8
108	Frequency of MELAS main mutation in a phenotype-targeted young ischemic stroke patient population. Journal of Neurology, 2016, 263, 257-262.	3.6	7

#	Article	IF	Citations
109	SNCA mutation p.Ala53Glu is derived from a common founder in the Finnish population. Neurobiology of Aging, 2017, 50, 168.e5-168.e8.	3.1	7
110	Defective mitochondrial ATPase due to rare mtDNA m.8969G>A mutationâ€"causing lactic acidosis, intellectual disability, and poor growth. Neurogenetics, 2018, 19, 49-53.	1.4	7
111	Quantitative solid-phase assay to measure deoxynucleoside triphosphate pools. Biology Methods and Protocols, 2018, 3, bpy011.	2.2	7
112	Quantitative Analysis of RNA Species by PCR and Solid-Phase Minisequencing. , 1998, 86, 121-132.		6
113	Genetic background of ataxia in children younger than 5 years in Finland. Neurology: Genetics, 2020, 6, e444.	1.9	6
114	Mitochondrial DNA Inheritance in Humans: Mix, Match, and Survival of the Fittest. Cell Metabolism, 2019, 30, 231-232.	16.2	5
115	Analysis of Nucleotide Sequence Variations by Solid-Phase Minisequencing. , 2003, 226, 361-366.		4
116	Generation and Characterization of Induced Pluripotent Stem Cells from Patients with mtDNA Mutations. Methods in Molecular Biology, 2015, 1353, 65-75.	0.9	4
117	Atomistic Molecular Dynamics Simulations of Mitochondrial DNA Polymerase Î³: Novel Mechanisms of Function and Pathogenesis. Biochemistry, 2017, 56, 1227-1238.	2.5	3
118	Atrial fibrillation is poorly tolerated by patients with hypertrophic concentric cardiomyopathy caused by mitochondrial tRNALeu (UUR) mutations. Neurology International, 2013, 3, .	0.5	2
119	Reply to: Proofreading deficiency in mitochondrial DNA polymerase does not affect total dNTP pools in mouse embryos. Nature Metabolism, 2020, 2, 676-677.	11.9	2
120	LCCS: A Lethal Motoneuron Disease of the Fetus Maps to Chromosome 9q34. Annals of the New York Academy of Sciences, 1998, 857, 260-262.	3.8	1
121	Modified Atkins diet modifies cardiopulmonary exercise characteristics and promotes hyperventilation in healthy subjects. Journal of Functional Foods, 2021, 81, 104459.	3.4	1
122	Krebs means cancer. Trends in Genetics, 2002, 18, 285-286.	6.7	0
123	Experimental models of muscle diseases. , 0, , 544-561.		0
124	Reply to â€˜Letter to Editor by Finsterer J and Zarrouk-Mahjoub S: Phenotypic manifestations of the m.8969G›A variantâ€™. Neurogenetics, 2018, 19, 133-134.	1.4	0
125	Diseases of DNA Polymerase Gamma. , 2019, , 113-124.		0
126	Whole-Cell and Mitochondrial dNTP Quantification from Cells and Tissues. Methods in Molecular Biology, 2021, 2276, 143-151.	0.9	0

#	Article	IF	Citations
127	The Increase of Mitochondrial DNA Copy Number Attenuates Eccentric Cardiac Remodeling In Volume Overload Model. FASEB Journal, 2013, 27, 1129.11.	0.5	0