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	Cost-effectiveness of whole-exome sequencing in progressive neurological disorders of children. European Journal of Paediatric Neurology, 2022, 36, 30-36.	1.6	12
2	Mosaic dysfunction of mitophagy in mitochondrial muscle disease. Cell Metabolism, 2022, 34, 197-208.e5.	16.2	35
3	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
4	Vegan diet in young children remodels metabolism and challenges the statuses of essential nutrients. EMBO Molecular Medicine, 2021, 13, e13492.	6.9	43
5	<scp>MED27</scp> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. Annals of Neurology, 2021, 89, 828-833.	5.3	14
6	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
7	Modified Atkins diet modifies cardiopulmonary exercise characteristics and promotes hyperventilation in healthy subjects. Journal of Functional Foods, 2021, 81, 104459.	3.4	1
8	Mitochondrial disease in adults: recent advances and future promise. Lancet Neurology, The, 2021, 20, 573-584.	10.2	96
9	IMPDH2: a new gene associated with dominant juvenile-onset dystonia-tremor disorder. European Journal of Human Genetics, 2021, 29, 1833-1837.	2.8	17
10	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
11	Whole-Cell and Mitochondrial dNTP Quantification from Cells and Tissues. Methods in Molecular Biology, 2021, 2276, 143-151.	0.9	0
12	SUCLA2 mutations cause global protein succinylation contributing to the pathomechanism of a hereditary mitochondrial disease. Nature Communications, 2020, 11, 5927.	12.8	35
13	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
14	Genetic background of ataxia in children younger than 5 years in Finland. Neurology: Genetics, 2020, 6, e444.	1.9	6
15	Niacin Cures Systemic NAD+ Deficiency and Improves Muscle Performance in Adult-Onset Mitochondrial Myopathy. Cell Metabolism, 2020, 31, 1078-1090.e5.	16.2	154
16	Using urine to diagnose largeâ€scale mtDNA deletions in adult patients. Annals of Clinical and Translational Neurology, 2020, 7, 1318-1326.	3.7	11
17	Integrative omics approaches provide biological and clinical insights: examples from mitochondrial diseases. Journal of Clinical Investigation, 2020, 130, 20-28.	8.2	39
18	Mitochondrial spongiotic brain disease: astrocytic stress and harmful rapamycin and ketosis effect. Life Science Alliance, 2020, 3, e202000797.	2.8	12

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19	Instability of the mitochondrial alanyl-tRNA synthetase underlies fatal infantile-onset cardiomyopathy. Human Molecular Genetics, 2019, 28, 258-268.	2.9	19
20	Mitochondrial DNA Inheritance in Humans: Mix, Match, and Survival of the Fittest. Cell Metabolism, 2019, 30, 231-232.	16.2	5
21	TFPa/HADHA is required for fatty acid beta-oxidation and cardiolipin re-modeling in human cardiomyocytes. Nature Communications, 2019, 10, 4671.	12.8	77
22	Disruption of the mouse Shmt2 gene confers embryonic anaemia via foetal liver-specific metabolomic disorders. Scientific Reports, 2019, 9, 16054.	3.3	8
23	Phenotypic effects of dietary stress in combination with a respiratory chain bypass in mice. Physiological Reports, 2019, 7, e14159.	1.7	8
24	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
25	Regulation of Mother-to-Offspring Transmission of mtDNA Heteroplasmy. Cell Metabolism, 2019, 30, 1120-1130.e5.	16.2	66
26	Diseases of DNA Polymerase Gamma. , 2019, , 113-124.		0
27	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
28	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
29	A urinary biosignature for mitochondrial myopathy, encephalopathy, lactic acidosis and stroke like episodes (MELAS). Mitochondrion, 2019, 45, 38-45.	3.4	16
30	Mitochondrial stress response triggered by defects in protein synthesis quality control. Life Science Alliance, 2019, 2, e201800219.	2.8	26
31	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
32	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
33	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
34	Loss of mtDNA activates astrocytes and leads to spongiotic encephalopathy. Nature Communications, 2018, 9, 70.	12.8	38
35	Retrospective natural history of thymidine kinase 2 deficiency. Journal of Medical Genetics, 2018, 55, 515-521.	3.2	73
36	Mitochondrial diseases: the contribution of organelle stress responses to pathology. Nature Reviews Molecular Cell Biology, 2018, 19, 77-92.	37.0	369

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37	Quantitative solid-phase assay to measure deoxynucleoside triphosphate pools. Biology Methods and Protocols, 2018, 3, bpy011.	2.2	7
38	Phosphorylation of Parkin at serine 65 is essential for its activationin vivo. Open Biology, 2018, 8, 180108.	3.6	81
39	Genetic Basis of Severe Childhood-OnsetÂCardiomyopathies. Journal of the American College of Cardiology, 2018, 72, 2324-2338.	2.8	97
40	RNA modification landscape of the human mitochondrial tRNALys regulates protein synthesis. Nature Communications, 2018, 9, 3966.	12.8	61
41	Metabolomes of mitochondrial diseases and inclusion body myositis patients: treatment targets and biomarkers. EMBO Molecular Medicine, 2018, 10, .	6.9	54
42	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
43	Atomistic Molecular Dynamics Simulations of Mitochondrial DNA Polymerase Î³: Novel Mechanisms of Function and Pathogenesis. Biochemistry, 2017, 56, 1227-1238.	2.5	3
44	mTORC1 Regulates Mitochondrial Integrated Stress Response and Mitochondrial Myopathy Progression. Cell Metabolism, 2017, 26, 419-428.e5.	16.2	291
45	Defective mitochondrial RNA processing due to PNPT1 variants causes Leigh syndrome. Human Molecular Genetics, 2017, 26, 3352-3361.	2.9	41
46	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
47	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
48	Modified Atkins diet induces subacute selective raggedâ€redâ€fiber lysis in mitochondrial myopathyÂpatients. EMBO Molecular Medicine, 2016, 8, 1234-1247.	6.9	56
49	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
50	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
51	Mitochondrial diseases. Nature Reviews Disease Primers, 2016, 2, 16080.	30.5	1,001
52	FGF21 is a biomarker for mitochondrial translation and mtDNA maintenance disorders. Neurology, 2016, 87, 2290-2299.	1.1	167
53	USF1 deficiency activates brown adipose tissue and improves cardiometabolic health. Science Translational Medicine, 2016, 8, 323ra13.	12.4	58
54	Mitochondrial DNA Replication Defects Disturb Cellular dNTP Pools and Remodel One-Carbon Metabolism. Cell Metabolism, 2016, 23, 635-648.	16.2	222

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55	Frequency of MELAS main mutation in a phenotype-targeted young ischemic stroke patient population. Journal of Neurology, 2016, 263, 257-262.	3.6	7
56	Mitochondrial roles in disease: a box full of surprises. EMBO Molecular Medicine, 2015, 7, 1245-1247.	6.9	11
57	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
58	Asymmetric rejuvenation. Nature, 2015, 521, 296-298.	27.8	8
59	Stem cells, mitochondria and aging. Biochimica Et Biophysica Acta - Bioenergetics, 2015, 1847, 1380-1386.	1.0	65
60	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
61	Selenoprotein biosynthesis defect causes progressive encephalopathy with elevated lactate. Neurology, 2015, 85, 306-315.	1.1	52
62	mtDNA Mutagenesis Disrupts Pluripotent Stem Cell Function by Altering Redox Signaling. Cell Reports, 2015, 11, 1614-1624.	6.4	66
63	Impaired Mitochondrial Biogenesis in Adipose Tissue in Acquired Obesity. Diabetes, 2015, 64, 3135-3145.	0.6	263
64	Generation and Characterization of Induced Pluripotent Stem Cells from Patients with mtDNA Mutations. Methods in Molecular Biology, 2015, 1353, 65-75.	0.9	4
65	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
66	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
67	Mitochondrial EFTs defects in juvenile-onset Leigh disease, ataxia, neuropathy, and optic atrophy. Neurology, 2014, 83, 743-751.	1.1	31
68	Effective treatment of mitochondrial myopathy by nicotinamide riboside, a vitamin <scp>B</scp>3. EMBO Molecular Medicine, 2014, 6, 721-731.	6.9	326
69	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
70	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
71	Mesencephalic complex I deficiency does not correlate with parkinsonism in mitochondrial DNA maintenance disorders. Brain, 2013, 136, 2379-2392.	7.6	41
72	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

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73	Atrial fibrillation is poorly tolerated by patients with hypertrophic concentric cardiomyopathy caused by mitochondrial tRNALeu (UUR) mutations. Neurology International, 2013, 3, .	0.5	2
74	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
75	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
76	The Increase of Mitochondrial DNA Copy Number Attenuates Eccentric Cardiac Remodeling In Volume Overload Model. FASEB Journal, 2013, 27, 1129.11.	0.5	0
77	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
78	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
79	Effect of bezafibrate treatment on late-onset mitochondrial myopathy in mice. Human Molecular Genetics, 2012, 21, 526-535.	2.9	125
80	Mechanisms of mitochondrial diseases. Annals of Medicine, 2012, 44, 41-59.	3.8	149
81	Mitochondria: In Sickness and in Health. Cell, 2012, 148, 1145-1159.	28.9	2,411
82	Somatic Progenitor Cell Vulnerability to Mitochondrial DNA Mutagenesis Underlies Progeroid Phenotypes in Polg Mutator Mice. Cell Metabolism, 2012, 15, 100-109.	16.2	213
83	Mitochondrial recessive ataxia syndrome mimicking dominant spinocerebellar ataxia. Journal of the Neurological Sciences, 2012, 315, 160-163.	0.6	18
84	Therapy for mitochondrial disorders: Little proof, high research activity, some promise. Seminars in Fetal and Neonatal Medicine, 2011, 16, 236-240.	2.3	41
85	Comparison of solution-based exome capture methods for next generation sequencing. Genome Biology, 2011, 12, R94.	9.6	237
86	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
87	Exome Sequencing Identifies Mitochondrial Alanyl-tRNA Synthetase Mutations in Infantile Mitochondrial Cardiomyopathy. American Journal of Human Genetics, 2011, 88, 635-642.	6.2	229
88	Biomarkers for mitochondrial respiratory chain disorders. Journal of Inherited Metabolic Disease, 2011, 34, 277-282.	3.6	41
89	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
90	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

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91	Ribonucleotide reductase is not limiting for mitochondrial DNA copy number in mice. Nucleic Acids Research, 2010, 38, 8208-8218.	14.5	28
92	Ketogenic diet slows down mitochondrial myopathy progression in mice. Human Molecular Genetics, 2010, 19, 1974-1984.	2.9	168
93	High mitochondrial DNA copy number has detrimental effects in mice. Human Molecular Genetics, 2010, 19, 2695-2705.	2.9	123
94	Mitochondrial myopathy induces a starvation-like response. Human Molecular Genetics, 2010, 19, 3948-3958.	2.9	249
95	Mitochondrial DNA depletion syndromes â€“ Many genes, common mechanisms. Neuromuscular Disorders, 2010, 20, 429-437.	0.6	169
96	Mouse models of mtDNA replication diseases. Methods, 2010, 51, 405-410.	3.8	12
97	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
98	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
99	Mouse models of mitochondrial DNA defects and their relevance for human disease. EMBO Reports, 2009, 10, 137-143.	4.5	84
100	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
101	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
102	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
103	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
104	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
105	Thymidine kinase 2 defects can cause multi-tissue mtDNA depletion syndrome. Brain, 2008, 131, 2841-2850.	7.6	73
106	Recessive Twinkle mutations in early onset encephalopathy with mtDNA depletion. Brain, 2007, 130, 3032-3040.	7.6	188
107	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
108	Phenotypic spectrum associated with mutations of the mitochondrial polymerase Â gene. Brain, 2006, 129, 1674-1684.	7.6	397

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109	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
110	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
111	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
112	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
113	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
114	Twinkle helicase is essential for mtDNA maintenance and regulates mtDNA copy number. Human Molecular Genetics, 2004, 13, 3219-3227.	2.9	202
115	Parkinsonism, premature menopause, and mitochondrial DNA polymerase Î³ mutations: clinical and molecular genetic study. Lancet, The, 2004, 364, 875-882.	13.7	538
116	Analysis of Nucleotide Sequence Variations by Solid-Phase Minisequencing. , 2003, 226, 361-366.		4
117	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
118	Krebs means cancer. Trends in Genetics, 2002, 18, 285-286.	6.7	0
119	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
120	Diseases caused by nuclear genes affecting mtDNA stability. American Journal of Medical Genetics Part A, 2001, 106, 53-61.	2.4	100
121	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
122	Quantitative Analysis of Human DNA Sequences by PCR and Solid-Phase Minisequencing. Molecular Biotechnology, 2000, 15, 123-132.	2.4	29
123	Role of Adenine Nucleotide Translocator 1 in mtDNA Maintenance. Science, 2000, 289, 782-785.	12.6	591
124	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
125	Quantitative Analysis of RNA Species by PCR and Solid-Phase Minisequencing. , 1998, 86, 121-132.		6
126	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

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127	Experimental models of muscle diseases. , 0, , 544-561.		0