

Jan Am Smeitink

List of Publications by Year in descending order

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Version: 2024-02-01

76
papers

5,900
citations

76196

40
h-index

74018

75
g-index

82
all docs

82
docs citations

82
times ranked

8016
citing authors

#	ARTICLE	IF	CITATIONS
1	Monogenic Mitochondrial Disorders. <i>New England Journal of Medicine</i> , 2012, 366, 1132-1141.	13.9	523
2	Mammalian Mitochondrial Complex I: Biogenesis, Regulation, and Reactive Oxygen Species Generation. <i>Antioxidants and Redox Signaling</i> , 2010, 12, 1431-1470.	2.5	353
3	Spectrophotometric Assay for Complex I of the Respiratory Chain in Tissue Samples and Cultured Fibroblasts. <i>Clinical Chemistry</i> , 2007, 53, 729-734.	1.5	340
4	A Post-Hoc Comparison of the Utility of Sanger Sequencing and Exome Sequencing for the Diagnosis of Heterogeneous Diseases. <i>Human Mutation</i> , 2013, 34, 1721-1726.	1.1	303
5	Smith-Lemli-Opitz Syndrome Is Caused by Mutations in the 7-Dehydrocholesterol Reductase Gene. <i>American Journal of Human Genetics</i> , 1998, 63, 329-338.	2.6	271
6	OXPHOS mutations and neurodegeneration. <i>EMBO Journal</i> , 2012, 32, 9-29.	3.5	214
7	Distinct Clinical Phenotypes Associated with a Mutation in the Mitochondrial Translation Elongation Factor EFTs. <i>American Journal of Human Genetics</i> , 2006, 79, 869-877.	2.6	169
8	Mutant Mitochondrial Elongation Factor G1 and Combined Oxidative Phosphorylation Deficiency. <i>New England Journal of Medicine</i> , 2004, 351, 2080-2086.	13.9	168
9	Mutations in NDUFAF3 (C3ORF60), Encoding an NDUFAF4 (C6ORF66)-Interacting Complex I Assembly Protein, Cause Fatal Neonatal Mitochondrial Disease. <i>American Journal of Human Genetics</i> , 2009, 84, 718-727.	2.6	155
10	Mutations in C12orf65 in Patients with Encephalomyopathy and a Mitochondrial Translation Defect. <i>American Journal of Human Genetics</i> , 2010, 87, 115-122.	2.6	144
11	Detection and manipulation of mitochondrial reactive oxygen species in mammalian cells. <i>Biochimica Et Biophysica Acta - Bioenergetics</i> , 2010, 1797, 1034-1044.	0.5	133
12	Identification of Mitochondrial Complex I Assembly Intermediates by Tracing Tagged NDUF53 Demonstrates the Entry Point of Mitochondrial Subunits. <i>Journal of Biological Chemistry</i> , 2007, 282, 7582-7590.	1.6	132
13	OXPHOS gene expression and control in mitochondrial disorders. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2009, 1792, 1113-1121.	1.8	128
14	Human mitochondrial complex I assembly: A dynamic and versatile process. <i>Biochimica Et Biophysica Acta - Bioenergetics</i> , 2007, 1767, 1215-1227.	0.5	125
15	Superoxide production is inversely related to complex I activity in inherited complex I deficiency. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2007, 1772, 373-381.	1.8	123
16	NDUFA2 Complex I Mutation Leads to Leigh Disease. <i>American Journal of Human Genetics</i> , 2008, 82, 1306-1315.	2.6	119
17	Impaired complex I assembly in a Leigh syndrome patient with a novel missense mutation in the ND6 gene. <i>Annals of Neurology</i> , 2003, 54, 665-669.	2.8	103
18	Inhibition of Mitochondrial Na ⁺ -Ca ²⁺ Exchange Restores Agonist-induced ATP Production and Ca ²⁺ Handling in Human Complex I Deficiency. <i>Journal of Biological Chemistry</i> , 2004, 279, 40328-40336.	1.6	101

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19	Simvastatin: a new therapeutic approach for Smith-Lemli-Opitz syndrome. <i>Journal of Lipid Research</i> , 2000, 41, 1339-1346.	2.0	98
20	Measurement of the Energy-Generating Capacity of Human Muscle Mitochondria: Diagnostic Procedure and Application to Human Pathology. <i>Clinical Chemistry</i> , 2006, 52, 860-871.	1.5	96
21	Bcl-2 prevents loss of mitochondria in CCCP-induced apoptosis. <i>Experimental Cell Research</i> , 2004, 299, 533-540.	1.2	95
22	Computer-assisted live cell analysis of mitochondrial membrane potential, morphology and calcium handling. <i>Methods</i> , 2008, 46, 304-311.	1.9	89
23	Mitochondrial disorders in children: toward development of small molecule treatment strategies. <i>EMBO Molecular Medicine</i> , 2016, 8, 311-327.	3.3	86
24	Mitochondrial hyperpolarization during chronic complex I inhibition is sustained by low activity of complex II, III, IV and V. <i>Biochimica Et Biophysica Acta - Bioenergetics</i> , 2014, 1837, 1247-1256.	0.5	81
25	A complex V ATP5A1 defect causes fatal neonatal mitochondrial encephalopathy. <i>Brain</i> , 2013, 136, 1544-1554.	3.7	80
26	Mutated ND2 impairs mitochondrial complex I assembly and leads to Leigh Syndrome. <i>Molecular Genetics and Metabolism</i> , 2007, 90, 10-14.	0.5	76
27	Mitochondrial and cytosolic thiol redox state are not detectably altered in isolated human NADH:ubiquinone oxidoreductase deficiency. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2007, 1772, 1041-1051.	1.8	69
28	Investigation of the complex I assembly chaperones B17.2L and NDUFAF1 in a cohort of CI deficient patients. <i>Molecular Genetics and Metabolism</i> , 2007, 91, 176-182.	0.5	68
29	Mitochondrial Retinal Dystrophy Associated with the m.3243A>G Mutation. <i>Ophthalmology</i> , 2013, 120, 2684-2696.	2.5	65
30	Trolox-Sensitive Reactive Oxygen Species Regulate Mitochondrial Morphology, Oxidative Phosphorylation and Cytosolic Calcium Handling in Healthy Cells. <i>Antioxidants and Redox Signaling</i> , 2012, 17, 1657-1669.	2.5	63
31	Mitochondrial Ca ²⁺ homeostasis in human NADH:ubiquinone oxidoreductase deficiency. <i>Cell Calcium</i> , 2008, 44, 123-133.	1.1	60
32	Metabolic consequences of NDUFS4 gene deletion in immortalized mouse embryonic fibroblasts. <i>Biochimica Et Biophysica Acta - Bioenergetics</i> , 2012, 1817, 1925-1936.	0.5	60
33	Subunits of Mitochondrial Complex I Exist as Part of Matrix- and Membrane-associated Subcomplexes in Living Cells. <i>Journal of Biological Chemistry</i> , 2008, 283, 34753-34761.	1.6	59
34	Novel mutations in the NDUFS1 gene cause low residual activities in human complex I deficiencies. <i>Molecular Genetics and Metabolism</i> , 2010, 100, 251-256.	0.5	53
35	SURFEIT-1 Gene Analysis and Two-Dimensional Blue Native Gel Electrophoresis in Cytochrome c Oxidase Deficiency. <i>Biochemical and Biophysical Research Communications</i> , 1999, 265, 339-344.	1.0	52
36	Mitigation of NADH: Ubiquinone oxidoreductase deficiency by chronic Trolox treatment. <i>Biochimica Et Biophysica Acta - Bioenergetics</i> , 2008, 1777, 853-859.	0.5	48

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37	Mitochondrial dynamics in human NADH:ubiquinone oxidoreductase deficiency. <i>International Journal of Biochemistry and Cell Biology</i> , 2009, 41, 1773-1782.	1.2	47
38	A novel mutation in COQ2 leading to fatal infantile multisystem disease. <i>Journal of the Neurological Sciences</i> , 2013, 326, 24-28.	0.3	45
39	Baculovirus complementation restores a novel <i>NDUFAF2</i> mutation causing complex I deficiency. <i>Human Mutation</i> , 2009, 30, E728-E736.	1.1	44
40	Complex I disorders: Causes, mechanisms, and development of treatment strategies at the cellular level. <i>Developmental Disabilities Research Reviews</i> , 2010, 16, 175-182.	2.9	43
41	Primary fibroblasts of <i>NDUFS4</i> ^{-/-} mice display increased ROS levels and aberrant mitochondrial morphology. <i>Mitochondrion</i> , 2013, 13, 436-443.	1.6	41
42	Cellular and animal models for mitochondrial complex I deficiency: A focus on the <i>NDUFS4</i> subunit. <i>IUBMB Life</i> , 2013, 65, 202-208.	1.5	40
43	Mitochondrial dysfunction in primary human fibroblasts triggers an adaptive cell survival program that requires AMPK- α . <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2015, 1852, 529-540.	1.8	40
44	Restoration of complex V deficiency caused by a novel deletion in the human <i>TMEM70</i> gene normalizes mitochondrial morphology. <i>Mitochondrion</i> , 2011, 11, 954-963.	1.6	39
45	Decreased agonist-stimulated mitochondrial ATP production caused by a pathological reduction in endoplasmic reticulum calcium content in human complex I deficiency. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2006, 1762, 115-123.	1.8	38
46	Pharmacological targeting of mitochondrial complex I deficiency: The cellular level and beyond. <i>Mitochondrion</i> , 2012, 12, 57-65.	1.6	38
47	Genetic defects in the oxidative phosphorylation (OXPHOS) system. <i>Expert Review of Molecular Diagnostics</i> , 2004, 4, 143-156.	1.5	37
48	Subunit-specific Incorporation Efficiency and Kinetics in Mitochondrial Complex I Homeostasis. <i>Journal of Biological Chemistry</i> , 2012, 287, 41851-41860.	1.6	34
49	Molecular base of biochemical complex I deficiency. <i>Mitochondrion</i> , 2012, 12, 520-532.	1.6	34
50	Developing outcome measures for pediatric mitochondrial disorders: Which complaints and limitations are most burdensome to patients and their parents?. <i>Mitochondrion</i> , 2013, 13, 15-24.	1.6	34
51	Obstetric complications in carriers of the m.3243A>G mutation, a retrospective cohort study on maternal and fetal outcome. <i>Mitochondrion</i> , 2015, 25, 98-103.	1.6	33
52	Toward high-content screening of mitochondrial morphology and membrane potential in living cells. <i>International Journal of Biochemistry and Cell Biology</i> , 2015, 63, 66-70.	1.2	30
53	Isolated Mitochondrial Complex I Deficiency: Explorative Data Analysis of Patient Cell Parameters. <i>Current Pharmaceutical Design</i> , 2011, 17, 4023-4033.	0.9	28
54	Partial complex I inhibition decreases mitochondrial motility and increases matrix protein diffusion as revealed by fluorescence correlation spectroscopy. <i>Biochimica Et Biophysica Acta - Bioenergetics</i> , 2007, 1767, 940-947.	0.5	27

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55	Mitochondrial DNA m.3242G>A mutation, an under diagnosed cause of hypertrophic cardiomyopathy and renal tubular dysfunction?. European Journal of Medical Genetics, 2012, 55, 552-556.	0.7	27
56	High-Throughput Assay to Measure Oxygen Consumption in Digitonin-Permeabilized Cells of Patients with Mitochondrial Disorders. Clinical Chemistry, 2010, 56, 424-431.	1.5	26
57	Bcl-2 protects against apoptosis induced by antimycin A and bongkreic acid without restoring cellular ATP levels. Biochimica Et Biophysica Acta - Bioenergetics, 2002, 1554, 57-65.	0.5	25
58	NDUFS4 deletion triggers loss of NDUFA12 in Ndufs4 mice and Leigh syndrome patients: A stabilizing role for NDUFAF2. Biochimica Et Biophysica Acta - Bioenergetics, 2020, 1861, 148213.	0.5	25
59	Mitochondrial oxidative phosphorylation system assembly in man: recent achievements. Current Opinion in Neurology, 2001, 14, 777-781.	1.8	24
60	Octa-arginine boosts the penetration of elastin-like polypeptide nanoparticles in 3D cancer models. European Journal of Pharmaceutics and Biopharmaceutics, 2019, 137, 175-184.	2.0	23
61	Towards the harmonization of outcome measures in children with mitochondrial disorders. Developmental Medicine and Child Neurology, 2013, 55, 698-706.	1.1	22
62	Three families with de novo m.3243A>G mutation. BBA Clinical, 2016, 6, 19-24.	4.1	22
63	Skeletal muscle mitochondria of NDUFS4 ^{+/+} mice display normal maximal pyruvate oxidation and ATP production. Biochimica Et Biophysica Acta - Bioenergetics, 2015, 1847, 526-533.	0.5	21
64	Variants in NGLY1 lead to intellectual disability, myoclonus epilepsy, sensorimotor axonal polyneuropathy and mitochondrial dysfunction. Clinical Genetics, 2020, 97, 556-566.	1.0	19
65	Effects of clofibrate and KH176 on life span and motor function in mitochondrial complex I-deficient mice. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2020, 1866, 165727.	1.8	15
66	Defective mitochondrial translation differently affects the live cell dynamics of complex I subunits. Biochimica Et Biophysica Acta - Bioenergetics, 2011, 1807, 1624-1633.	0.5	13
67	A Heterozygous NDUFV1 Variant Aggravates Mitochondrial Complex I Deficiency in a Family with a Homoplasmic ND1 Variant. Journal of Pediatrics, 2018, 196, 309-313.e3.	0.9	13
68	New perspectives on the assembly process of mitochondrial respiratory chain complex cytochrome c oxidase. Mitochondrion, 2002, 2, 117-128.	1.6	10
69	Cognitive functioning and mental health in mitochondrial disease: A systematic scoping review. Neuroscience and Biobehavioral Reviews, 2021, 125, 57-77.	2.9	10
70	A novel mitochondrial DNA m.7507A>G mutation is only pathogenic at high levels of heteroplasmy. Neuromuscular Disorders, 2015, 25, 262-267.	0.3	9
71	Stimulation of cholesterol biosynthesis in mitochondrial complex I-deficiency lowers reductive stress and improves motor function and survival in mice. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2021, 1867, 166062.	1.8	7
72	Assisted 6-minute cycling test: An exploratory study in children. Muscle and Nerve, 2016, 54, 232-238.	1.0	5

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73	The decylTPP mitochondria-targeting moiety lowers electron transport chain supercomplex levels in primary human skin fibroblasts. <i>Free Radical Biology and Medicine</i> , 2022, 188, 434-446.	1.3	5
74	Mitochondrial DNA replication and OXPHOS gene transcription show varied responsiveness to Rieske protein knockdown in 143B cells. <i>Biochimie</i> , 2011, 93, 758-765.	1.3	3
75	New approaches to diagnosing mitochondrial abnormalities: Taking the next step. <i>Journal of Pediatric Biochemistry</i> , 2015, 02, 205-212.	0.2	0
76	Restoring cellular NAD(P)H levels by PPAR α and LXR α stimulation to improve mitochondrial complex I deficiency. <i>Life Sciences</i> , 2022, 300, 120571.	2.0	0