Jan Am Smeitink

List of Publications by Year
in descending order

76 papers	5,900 citations	76326 40 h-index	74163 75 g-index
82 all docs	82 docs citations	82 times ranked	8016 citing authors

#	Article	IF	Citations
1	Monogenic Mitochondrial Disorders. New England Journal of Medicine, 2012, 366, 1132-1141.	27.0	523
2	Mammalian Mitochondrial Complex I: Biogenesis, Regulation, and Reactive Oxygen Species Generation. Antioxidants and Redox Signaling, 2010, 12, 1431-1470.	5.4	353
3	Spectrophotometric Assay for Complex I of the Respiratory Chain in Tissue Samples and Cultured Fibroblasts. Clinical Chemistry, 2007, 53, 729-734.	3.2	340
4	A Post-Hoc Comparison of the Utility of Sanger Sequencing and Exome Sequencing for the Diagnosis of Heterogeneous Diseases. Human Mutation, 2013, 34, 1721-1726.	2.5	303
5	Smith-Lemli-Opitz Syndrome Is Caused by Mutations in the 7-Dehydrocholesterol Reductase Gene. American Journal of Human Genetics, 1998, 63, 329-338.	6.2	271
6	OXPHOS mutations and neurodegeneration. EMBO Journal, 2012, 32, 9-29.	7.8	214
7	Distinct Clinical Phenotypes Associated with a Mutation in the Mitochondrial Translation Elongation Factor EFTs. American Journal of Human Genetics, 2006, 79, 869-877.	6.2	169
8	Mutant Mitochondrial Elongation Factor G1 and Combined Oxidative Phosphorylation Deficiency. New England Journal of Medicine, 2004, 351, 2080-2086.	27.0	168
9	Mutations in NDUFAF3 (C3ORF60), Encoding an NDUFAF4 (C6ORF66)-Interacting Complex I Assembly Protein, Cause Fatal Neonatal Mitochondrial Disease. American Journal of Human Genetics, 2009, 84, 718-727.	6.2	155
10	Mutations in C12orf65 in Patients with Encephalomyopathy and a Mitochondrial Translation Defect. American Journal of Human Genetics, 2010, 87, 115-122.	6.2	144
11	Detection and manipulation of mitochondrial reactive oxygen species in mammalian cells. Biochimica Et Biophysica Acta - Bioenergetics, 2010, 1797, 1034-1044.	1.0	133
12	Identification of Mitochondrial Complex I Assembly Intermediates by Tracing Tagged NDUFS3 Demonstrates the Entry Point of Mitochondrial Subunits. Journal of Biological Chemistry, 2007, 282, 7582-7590.	3.4	132
13	OXPHOS gene expression and control in mitochondrial disorders. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2009, 1792, 1113-1121.	3.8	128
14	Human mitochondrial complex I assembly: A dynamic and versatile process. Biochimica Et Biophysica Acta - Bioenergetics, 2007, 1767, 1215-1227.	1.0	125
15	Superoxide production is inversely related to complex I activity in inherited complex I deficiency. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2007, 1772, 373-381.	3.8	123
16	NDUFA2 Complex I Mutation Leads to Leigh Disease. American Journal of Human Genetics, 2008, 82, 1306-1315.	6.2	119
17	Impaired complex I assembly in a Leigh syndrome patient with a novel missense mutation in the ND6 gene. Annals of Neurology, 2003, 54, 665-669.	5.3	103
18	Inhibition of Mitochondrial Na+-Ca2+ Exchange Restores Agonist-induced ATP Production and Ca2+ Handling in Human Complex I Deficiency. Journal of Biological Chemistry, 2004, 279, 40328-40336.	3.4	101

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19	Simvastatin: a new therapeutic approach for Smith-Lemli-Opitz syndrome. Journal of Lipid Research, 2000, 41, 1339-1346.	4.2	98
20	Measurement of the Energy-Generating Capacity of Human Muscle Mitochondria: Diagnostic Procedure and Application to Human Pathology. Clinical Chemistry, 2006, 52, 860-871.	3.2	96
21	Bcl-2 prevents loss of mitochondria in CCCP-induced apoptosis. Experimental Cell Research, 2004, 299, 533-540.	2.6	95
22	Computer-assisted live cell analysis of mitochondrial membrane potential, morphology and calcium handling. Methods, 2008, 46, 304-311.	3.8	89
23	Mitochondrial disorders in children: toward development of smallâ€molecule treatment strategies. EMBO Molecular Medicine, 2016, 8, 311-327.	6.9	86
24	Mitochondrial hyperpolarization during chronic complex I inhibition is sustained by low activity of complex II, III, IV and V. Biochimica Et Biophysica Acta - Bioenergetics, 2014, 1837, 1247-1256.	1.0	81
25	A complex V ATP5A1 defect causes fatal neonatal mitochondrial encephalopathy. Brain, 2013, 136, 1544-1554.	7.6	80
26	Mutated ND2 impairs mitochondrial complex I assembly and leads to Leigh Syndrome. Molecular Genetics and Metabolism, 2007, 90, 10-14.	1.1	76
27	Mitochondrial and cytosolic thiol redox state are not detectably altered in isolated human NADH:ubiquinone oxidoreductase deficiency. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2007, 1772, 1041-1051.	3.8	69
28	Investigation of the complex I assembly chaperones B17.2L and NDUFAF1 in a cohort of CI deficient patients. Molecular Genetics and Metabolism, 2007, 91, 176-182.	1.1	68
29	Mitochondrial Retinal Dystrophy Associated with the m.3243A>G Mutation. Ophthalmology, 2013, 120, 2684-2696.	5.2	65
30	Trolox-Sensitive Reactive Oxygen Species Regulate Mitochondrial Morphology, Oxidative Phosphorylation and Cytosolic Calcium Handling in Healthy Cells. Antioxidants and Redox Signaling, 2012, 17, 1657-1669.	5.4	63
31	Mitochondrial Ca2+ homeostasis in human NADH:ubiquinone oxidoreductase deficiencyâ˜†. Cell Calcium, 2008, 44, 123-133.	2.4	60
32	Metabolic consequences of NDUFS4 gene deletion in immortalized mouse embryonic fibroblasts. Biochimica Et Biophysica Acta - Bioenergetics, 2012, 1817, 1925-1936.	1.0	60
33	Subunits of Mitochondrial Complex I Exist as Part of Matrix- and Membrane-associated Subcomplexes in Living Cells. Journal of Biological Chemistry, 2008, 283, 34753-34761.	3.4	59
34	Novel mutations in the NDUFS1 gene cause low residual activities in human complex I deficiencies. Molecular Genetics and Metabolism, 2010, 100, 251-256.	1.1	53
35	SURFEIT-1 Gene Analysis and Two-Dimensional Blue Native Gel Electrophoresis in Cytochrome c Oxidase Deficiency. Biochemical and Biophysical Research Communications, 1999, 265, 339-344.	2.1	52
36	Mitigation of NADH: Ubiquinone oxidoreductase deficiency by chronic Trolox treatment. Biochimica Et Biophysica Acta - Bioenergetics, 2008, 1777, 853-859.	1.0	48

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37	Mitochondrial dynamics in human NADH:ubiquinone oxidoreductase deficiency. International Journal of Biochemistry and Cell Biology, 2009, 41, 1773-1782.	2.8	47
38	A novel mutation in COQ2 leading to fatal infantile multisystem disease. Journal of the Neurological Sciences, 2013, 326, 24-28.	0.6	45
39	Baculovirus complementation restores a novelNDUFAF2mutation causing complex I deficiency. Human Mutation, 2009, 30, E728-E736.	2.5	44
40	Complex I disorders: Causes, mechanisms, and development of treatment strategies at the cellular level. Developmental Disabilities Research Reviews, 2010, 16, 175-182.	2.9	43
41	Primary fibroblasts of NDUFS4âˆ’/âˆ’ mice display increased ROS levels and aberrant mitochondrial morphology. Mitochondrion, 2013, 13, 436-443.	3.4	41
42	Cellular and animal models for mitochondrial complex I deficiency: A focus on the NDUFS4 subunit. IUBMB Life, 2013, 65, 202-208.	3.4	40
43	Mitochondrial dysfunction in primary human fibroblasts triggers an adaptive cell survival program that requires AMPK-Î±. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2015, 1852, 529-540.	3.8	40
44	Restoration of complex V deficiency caused by a novel deletion in the human TMEM70 gene normalizes mitochondrial morphology. Mitochondrion, 2011, 11, 954-963.	3.4	39
45	Decreased agonist-stimulated mitochondrial ATP production caused by a pathological reduction in endoplasmic reticulum calcium content in human complex I deficiency. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2006, 1762, 115-123.	3.8	38
46	Pharmacological targeting of mitochondrial complex I deficiency: The cellular level and beyond. Mitochondrion, 2012, 12, 57-65.	3.4	38
47	Genetic defects in the oxidative phosphorylation (OXPHOS) system. Expert Review of Molecular Diagnostics, 2004, 4, 143-156.	3.1	37
48	Subunit-specific Incorporation Efficiency and Kinetics in Mitochondrial Complex I Homeostasis. Journal of Biological Chemistry, 2012, 287, 41851-41860.	3.4	34
49	Molecular base of biochemical complex I deficiency. Mitochondrion, 2012, 12, 520-532.	3.4	34
50	Developing outcome measures for pediatric mitochondrial disorders: Which complaints and limitations are most burdensome to patients and their parents?. Mitochondrion, 2013, 13, 15-24.	3.4	34
51	Obstetric complications in carriers of the m.3243A>G mutation, a retrospective cohort study on maternal and fetal outcome. Mitochondrion, 2015, 25, 98-103.	3.4	33
52	Toward high-content screening of mitochondrial morphology and membrane potential in living cells. International Journal of Biochemistry and Cell Biology, 2015, 63, 66-70.	2.8	30
53	Isolated Mitochondrial Complex I Deficiency: Explorative Data Analysis of Patient Cell Parameters. Current Pharmaceutical Design, 2011, 17, 4023-4033.	1.9	28
54	Partial complex I inhibition decreases mitochondrial motility and increases matrix protein diffusion as revealed by fluorescence correlation spectroscopy. Biochimica Et Biophysica Acta - Bioenergetics, 2007, 1767, 940-947.	1.0	27

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55	Mitochondrial DNA m.3242GÂ>ÂA mutation, an under diagnosed cause of hypertrophic cardiomyopathy and renal tubular dysfunction?. European Journal of Medical Genetics, 2012, 55, 552-556.	1.3	27
56	High-Throughput Assay to Measure Oxygen Consumption in Digitonin-Permeabilized Cells of Patients with Mitochondrial Disorders. Clinical Chemistry, 2010, 56, 424-431.	3.2	26
57	Bcl-2 protects against apoptosis induced by antimycin A and bongkrekic acid without restoring cellular ATP levels. Biochimica Et Biophysica Acta - Bioenergetics, 2002, 1554, 57-65.	1.0	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.	1.0	25
59	Mitochondrial oxidative phosphorylation system assembly in man: recent achievements. Current Opinion in Neurology, 2001, 14, 777-781.	3.6	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.	4.3	23
61	Towards the harmonization of outcome measures in children with mitochondrial disorders. Developmental Medicine and Child Neurology, 2013, 55, 698-706.	2.1	22
62	Three families with â€˜de novoâ€™ m.3243A>G mutation. BBA Clinical, 2016, 6, 19-24.	4.1	22
63	Skeletal muscle mitochondria of NDUFS4âˆ’/âˆ’ mice display normal maximal pyruvate oxidation and ATP production. Biochimica Et Biophysica Acta - Bioenergetics, 2015, 1847, 526-533.	1.0	21
64	Variants in NGLY1 lead to intellectual disability, myoclonus epilepsy, sensorimotor axonal polyneuropathy and mitochondrial dysfunction. Clinical Genetics, 2020, 97, 556-566.	2.0	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.	3.8	15
66	Defective mitochondrial translation differently affects the live cell dynamics of complex I subunits. Biochimica Et Biophysica Acta - Bioenergetics, 2011, 1807, 1624-1633.	1.0	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.	1.8	13
68	New perspectives on the assembly process of mitochondrial respiratory chain complex cytochrome c oxidase. Mitochondrion, 2002, 2, 117-128.	3.4	10
69	Cognitive functioning and mental health in mitochondrial disease: A systematic scoping review. Neuroscience and Biobehavioral Reviews, 2021, 125, 57-77.	6.1	10
70	A novel mitochondrial DNA m.7507A>G mutation is only pathogenic at high levels of heteroplasmy. Neuromuscular Disorders, 2015, 25, 262-267.	0.6	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.	3.8	7
72	Assisted 6-minute cycling test: An exploratory study in children. Muscle and Nerve, 2016, 54, 232-238.	2.2	5

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73	The decylTPP mitochondria-targeting moiety lowers electron transport chain supercomplex levels in primary human skin fibroblasts. Free Radical Biology and Medicine, 2022, 188, 434-446.	2.9	5
74	Mitochondrial DNA replication and OXPHOS gene transcription show varied responsiveness to Rieske protein knockdown in 143B cells. Biochimie, 2011, 93, 758-765.	2.6	3
75	New approaches to diagnosing mitochondrial abnormalities: Taking the next step. Journal of Pediatric Biochemistry, 2015, 02, 205-212.	0.2	0
76	Restoring cellular NAD(P)H levels by PPARÎ± and LXRÎ± stimulation to improve mitochondrial complex I deficiency. Life Sciences, 2022, 300, 120571.	4.3	0