

# Jan W Taanman

## List of Publications by Year in descending order

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119  
papers

8,718  
citations

53794

45  
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43889

91  
g-index

126  
all docs

126  
docs citations

126  
times ranked

10069  
citing authors

#	ARTICLE	IF	CITATIONS
1	Ambroxol reverses tau and $\beta$ -synuclein accumulation in a cholinergic N370S <i>GBA1</i> mutation model. <i>Human Molecular Genetics</i> , 2022, 31, 2396-2405.	2.9	10
2	Sirtuin 5 depletion impairs mitochondrial function in human proximal tubular epithelial cells. <i>Scientific Reports</i> , 2021, 11, 15510.	3.3	18
3	Mitochondrial respiratory chain and Krebs cycle enzyme function in human donor livers subjected to end-ischaemic hypothermic machine perfusion. <i>PLoS ONE</i> , 2021, 16, e0257783.	2.5	0
4	The PINK1/Parkin mitophagy signalling pathway is not functional in peripheral blood mononuclear cells. <i>PLoS ONE</i> , 2021, 16, e0259903.	2.5	8
5	Mitochondria as target to inhibit proliferation and induce apoptosis of cancer cells: the effects of doxycycline and gemcitabine. <i>Scientific Reports</i> , 2020, 10, 4363.	3.3	59
6	Mitochondrial DNA: Structure, Genetics, Replication and Defects. , 2019, , 127-152.		0
7	Huntingtin Aggregates and Mitochondrial Pathology in Skeletal Muscle but not Heart of Late-Stage R6/2 Mice. <i>Journal of Huntington's Disease</i> , 2019, 8, 145-159.	1.9	20
8	NDUFA4 (Renamed COXFA4) Is a Cytochrome-c Oxidase Subunit. <i>Trends in Endocrinology and Metabolism</i> , 2018, 29, 452-454.	7.1	44
9	Somatic copy number gains of $\beta$ -synuclein (SNCA) in Parkinson's disease and multiple system atrophy brains. <i>Brain</i> , 2018, 141, 2419-2431.	7.6	63
10	Mitochondria as oncotarget: a comparison between the tetracycline analogs doxycycline and COL-3. <i>Oncotarget</i> , 2018, 9, 33818-33831.	1.8	18
11	Impact of treprostinil on dynamin-related protein 1 (DRP1) and mitochondrial fragmentation in pulmonary arterial hypertension (PAH).. , 2018, , .		1
12	A Human Neural Crest Stem Cell-Derived Dopaminergic Neuronal Model Recapitulates Biochemical Abnormalities in <i>GBA1</i> Mutation Carriers. <i>Stem Cell Reports</i> , 2017, 8, 728-742.	4.8	57
13	Clinicopathologic and molecular spectrum of <i>RNASEH1</i> -related mitochondrial disease. <i>Neurology: Genetics</i> , 2017, 3, e149.	1.9	19
14	The clinical spectrum and natural history of early-onset diseases due to DNA polymerase gamma mutations. <i>Genetics in Medicine</i> , 2017, 19, 1217-1225.	2.4	45
15	Mitochondrial cristae remodelling is associated with disrupted OPA1 oligomerisation in the Huntington's disease R6/2 fragment model. <i>Experimental Neurology</i> , 2017, 288, 167-175.	4.1	27
16	Loss of PINK1 or Parkin Function Results in a Progressive Loss of Mitochondrial Function. , 2017, , 187-209.		1
17	DNA isolation protocol effects on nuclear DNA analysis by microarrays, droplet digital PCR, and whole genome sequencing, and on mitochondrial DNA copy number estimation. <i>PLoS ONE</i> , 2017, 12, e0180467.	2.5	27
18	Subunit composition of respiratory chain complex 1 and its responses to oxygen in mitochondria from human donor livers. <i>BMC Research Notes</i> , 2017, 10, 547.	1.4	1

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19	A Clinical, Neuropathological and Genetic Study of Homozygous A467T POLG-Related Mitochondrial Disease. PLoS ONE, 2016, 11, e0145500.	2.5	36
20	A LON-ClpP Proteolytic Axis Degrades Complex I to Extinguish ROS Production in Depolarized Mitochondria. Cell Reports, 2016, 17, 2522-2531.	6.4	81
21	TRNT1 deficiency: clinical, biochemical and molecular genetic features. Orphanet Journal of Rare Diseases, 2016, 11, 90.	2.7	64
22	B25â€¦Mitochondrial fission and fusion in skeletal muscle from HD patients and zQ175 mice. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, A17.3-A18.	1.9	0
23	Selective striatal mtDNA depletion in end-stage Huntington's disease R6/2 mice. Experimental Neurology, 2015, 266, 22-29.	4.1	19
24	Clonal Expansion of T Cells in Abdominal Aortic Aneurysm: A Role for Doxycycline as Drug of Choice?. International Journal of Molecular Sciences, 2015, 16, 11178-11195.	4.1	12
25	Signal transducer and activator of transcription 2 deficiency is a novel disorder of mitochondrial fission. Brain, 2015, 138, 2834-2846.	7.6	78
26	D03 Mitochondrial Biogenesis, And Respiratory Chain Assembly And Function, In Skeletal Muscle Of The R6/2 Mouse Model And Human Huntington's Disease. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, A32-A32.	1.9	0
27	Comment on "Aneurysmal Lesions of Patients with Abdominal Aortic Aneurysm Contain Clonally Expanded T Cells". Journal of Immunology, 2014, 193, 2041.1-2041.	0.8	2
28	B34 Mitochondrial Biogenesis, And Respiratory Chain Assembly And Function, In Heart Of R6/2 Mouse Model Of Huntington's Disease. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, A21-A21.	1.9	0
29	NDUFA4 Mutations Underlie Dysfunction of a Cytochrome c Oxidase Subunit Linked to Human Neurological Disease. Cell Reports, 2013, 3, 1795-1805.	6.4	104
30	Pathogenic Parkinson's disease mutations across the functional domains of LRRK2 alter the autophagic/lysosomal response to starvation. Biochemical and Biophysical Research Communications, 2013, 441, 862-866.	2.1	79
31	HIBCH mutations can cause Leigh-like disease with combined deficiency of multiple mitochondrial respiratory chain enzymes and pyruvate dehydrogenase. Orphanet Journal of Rare Diseases, 2013, 8, 188.	2.7	70
32	NDUFA4 Mutations Underlie Dysfunction of a Cytochrome c Oxidase Subunit Linked to Human Neurological Disease. Cell Reports, 2013, 4, 402.	6.4	0
33	Glucosylceramidase degradation in fibroblasts carrying bi-allelic Parkin mutations. Molecular Genetics and Metabolism, 2013, 109, 402-403.	1.1	5
34	COX10 Mutations Resulting in Complex Multisystem Mitochondrial Disease That Remains Stable Into Adulthood. JAMA Neurology, 2013, 70, 1556-61.	9.0	27
35	Mitofusin 1 and mitofusin 2 are ubiquitinated in a PINK1/parkin-dependent manner upon induction of mitophagy. Human Molecular Genetics, 2013, 22, 1697-1697.	2.9	4
36	Creation of an Open-Access, Mutation-Defined Fibroblast Resource for Neurological Disease Research. PLoS ONE, 2012, 7, e43099.	2.5	44

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37	Kearns-Sayre syndrome caused by defective R1/p53R2 assembly. <i>Journal of Medical Genetics</i> , 2011, 48, 610-617.	3.2	49
38	P47 Mutations in the novel chaperone FOXRED1 cause mitochondrial complex I deficiency. <i>Neuromuscular Disorders</i> , 2011, 21, S19.	0.6	0
39	P58 Mitochondrial respiratory chain enzyme deficiency expressed during muscle development. <i>Neuromuscular Disorders</i> , 2011, 21, S23.	0.6	0
40	Pathogenic LRRK2 Mutations Do Not Alter Gene Expression in Cell Model Systems or Human Brain Tissue. <i>PLoS ONE</i> , 2011, 6, e22489.	2.5	27
41	Influence of zinc and zinc chelator on HT-29 colorectal cell line. <i>BioMetals</i> , 2011, 24, 143-151.	4.1	10
42	Mutations in the mitochondrial complex I assembly factor NDUF1L cause fatal infantile hypertrophic cardiomyopathy. <i>Journal of Medical Genetics</i> , 2011, 48, 691-697.	3.2	64
43	14 Mitochondrial cardiomyopathy caused by defective assembly of respiratory chain complex I. <i>Heart</i> , 2011, 97, e8-e8.	2.9	0
44	Parkinson's disease induced pluripotent stem cells with triplication of the $\alpha$ -synuclein locus. <i>Nature Communications</i> , 2011, 2, 440.	12.8	406
45	Mitochondrial single-stranded DNA binding protein is required for maintenance of mitochondrial DNA and 7S DNA but is not required for mitochondrial nucleoid organisation. <i>Biochimica Et Biophysica Acta - Molecular Cell Research</i> , 2010, 1803, 931-939.	4.1	64
46	Lowering the apoptotic threshold in colorectal cancer cells by targeting mitochondria. <i>Cancer Cell International</i> , 2010, 10, 31.	4.1	10
47	Mitofusin 1 and mitofusin 2 are ubiquitinated in a PINK1/parkin-dependent manner upon induction of mitophagy. <i>Human Molecular Genetics</i> , 2010, 19, 4861-4870.	2.9	795
48	FOXRED1, encoding an FAD-dependent oxidoreductase complex-I-specific molecular chaperone, is mutated in infantile-onset mitochondrial encephalopathy. <i>Human Molecular Genetics</i> , 2010, 19, 4837-4847.	2.9	79
49	POG01 Anti-thymidine phosphorylase antibodies in the diagnosis of mitochondrial neurogastrointestinal encephalomyopathy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2010, 81, e48-e48.	1.9	0
50	The diagnosis of inherited metabolic diseases by microarray gene expression profiling. <i>Orphanet Journal of Rare Diseases</i> , 2010, 5, 34.	2.7	3
51	Intracellular oxygenation and cytochrome oxidase C activity in ischemic preconditioning of steatotic rabbit liver. <i>American Journal of Surgery</i> , 2010, 200, 507-518.	1.8	10
52	Measurement of kinetic parameters of human platelet DNA polymerase $\beta$ . <i>Methods</i> , 2010, 51, 374-378.	3.8	2
53	Analysis of mutant DNA polymerase $\beta$ in patients with mitochondrial DNA depletion. <i>Human Mutation</i> , 2009, 30, 248-254.	2.5	52
54	Status epilepticus in children with Alpers' disease caused by <i>POLG1</i> mutations: EEG and MRI features. <i>Epilepsia</i> , 2009, 50, 1596-1607.	5.1	141

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55	Increased sensitivity of myoblasts to oxidative stress in amyotrophic lateral sclerosis peripheral tissues. <i>Experimental Neurology</i> , 2009, 218, 92-97.	4.1	16
56	Differential effects of PINK1 nonsense and missense mutations on mitochondrial function and morphology. <i>Experimental Neurology</i> , 2009, 219, 266-273.	4.1	93
57	Collated mutations in mitochondrial DNA (mtDNA) depletion syndrome (excluding the mitochondrial) Tj ETQq1 1 0.784314 rgBT /Ove 1109-1112.	3.8	41
58	Characterization of a novel TYMP splice site mutation associated with mitochondrial neurogastrointestinal encephalomyopathy (MNGIE). <i>Neuromuscular Disorders</i> , 2009, 19, 151-154.	0.6	29
59	Does Doxycycline work in synergy with cisplatin and oxaliplatin in colorectal cancer?. <i>World Journal of Surgical Oncology</i> , 2009, 7, 2.	1.9	16
60	Silencing of PINK1 Expression Affects Mitochondrial DNA and Oxidative Phosphorylation in DOPAMINERGIC Cells. <i>PLoS ONE</i> , 2009, 4, e4756.	2.5	173
61	Influence of mitochondrial DNA level on cellular energy metabolism: implications for mitochondrial diseases. <i>Journal of Bioenergetics and Biomembranes</i> , 2008, 40, 59-67.	2.3	57
62	Kinetic properties of mutant deoxyguanosine kinase in a case of reversible hepatic mtDNA depletion. <i>Biochemical Journal</i> , 2007, 402, 377-385.	3.7	39
63	Myoclonusâ€“dystonia syndrome with severe depression is caused by an exon-skipping mutation in the $\epsilon$ -sarcoglycan gene. <i>Movement Disorders</i> , 2007, 22, 1173-1175.	3.9	41
64	Relapsing neuropathy in an 18-year-old woman. <i>Lancet Neurology</i> , The, 2007, 6, 192-198.	10.2	4
65	Phenotypic variability of mitochondrial disease caused by a nuclear mutation in complex II. <i>Molecular Genetics and Metabolism</i> , 2006, 89, 214-221.	1.1	42
66	Dominant inheritance of premature ovarian failure associated with mutant mitochondrial DNA polymerase gamma. <i>Human Reproduction</i> , 2006, 21, 2467-2473.	0.9	153
67	Analysis of COX2 mutants reveals cytochrome oxidase subassemblies in yeast. <i>Biochemical Journal</i> , 2005, 390, 703-708.	3.7	31
68	Mutant torsinA, which causes early-onset primary torsion dystonia, is redistributed to membranous structures enriched in vesicular monoamine transporter in cultured human SH-SY5Y cells. <i>Movement Disorders</i> , 2005, 20, 432-440.	3.9	48
69	Assessment of the significance of mitochondrial DNA damage by chemotherapeutic agents. <i>International Journal of Oncology</i> , 2005, 27, 337.	3.3	2
70	Clinical, biochemical and morphological features of hepatocerebral syndrome with mitochondrial DNA depletion due to deoxyguanosine kinase deficiency. <i>Journal of Hepatology</i> , 2005, 43, 333-341.	3.7	75
71	Analysis of the trinucleotide CAG repeat from the DNA polymerase $\beta$ gene (POLG) in patients with Parkinson's disease. <i>Neuroscience Letters</i> , 2005, 376, 56-59.	2.1	39
72	The Human Mitochondrial Genome. <i>Oxidative Stress and Disease</i> , 2005, , 95-246.	0.3	1

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73	Cytochrome c Oxidase Subassemblies in Fibroblast Cultures from Patients Carrying Mutations in COX10, SCO1, or SURF1. <i>Journal of Biological Chemistry</i> , 2004, 279, 7462-7469.	3.4	118
74	Replication of mitochondrial DNA occurs throughout the mitochondria of cultured human cells. <i>Experimental Cell Research</i> , 2003, 289, 133-142.	2.6	86
75	Mitochondrial DNA depletion can be prevented by dGMP and dAMP supplementation in a resting culture of deoxyguanosine kinase-deficient fibroblasts. <i>Human Molecular Genetics</i> , 2003, 12, 1839-1845.	2.9	78
76	Chapter 1 Structure and Function of the Mitochondrial Oxidative Phosphorylation System. <i>Blue Books of Practical Neurology</i> , 2002, , 1-34.	0.1	3
77	Mutations of cytochrome c oxidase subunits 1 and 3 in <i>Saccharomyces cerevisiae</i> : assembly defect and compensation. <i>Biochimica Et Biophysica Acta - Bioenergetics</i> , 2002, 1554, 101-107.	1.0	21
78	Depletion of mitochondrial DNA in the liver of an infant with neonatal giant cell hepatitis. <i>Human Pathology</i> , 2002, 33, 247-253.	2.0	42
79	A novel mutation in the deoxyguanosine kinase gene causing depletion of mitochondrial DNA. <i>Annals of Neurology</i> , 2002, 52, 237-239.	5.3	44
80	Diagnostic Value of Succinate Ubiquinone Reductase Activity in the Identification of Patients with Mitochondrial DNA Depletion. <i>Journal of Inherited Metabolic Disease</i> , 2002, 25, 7-16.	3.6	26
81	A Novel Mutation in SURF1 Causes Skipping of Exon 8 in a Patient with Cytochrome c Oxidase-Deficient Leigh Syndrome and Hypertrichosis. <i>Molecular Genetics and Metabolism</i> , 2001, 73, 340-343.	1.1	22
82	Assembly of cytochrome c oxidase: what can we learn from patients with cytochrome c oxidase deficiency?. <i>Biochemical Society Transactions</i> , 2001, 29, 446-451.	3.4	30
83	Immunological Phenotyping of Fibroblast Cultures from Patients with a Mitochondrial Respiratory Chain Deficit. <i>Laboratory Investigation</i> , 2001, 81, 1069-1077.	3.7	17
84	A mutant mitochondrial respiratory chain assembly protein causes complex III deficiency in patients with tubulopathy, encephalopathy and liver failure. <i>Nature Genetics</i> , 2001, 29, 57-60.	21.4	297
85	A nuclear modifier for a mitochondrial DNA disorder. <i>Trends in Genetics</i> , 2001, 17, 609-611.	6.7	7
86	Mitochondrial Dysfunction in Congenital Nephrotic Syndrome. <i>Laboratory Investigation</i> , 2000, 80, 1227-1232.	3.7	25
87	A novel frameshift mutation of the mtDNA COIII gene leads to impaired assembly of cytochrome c oxidase in a patient affected by Leigh-like syndrome. <i>Human Molecular Genetics</i> , 2000, 9, 2733-2742.	2.9	109
88	A mutation in the human heme A:farnesyltransferase gene (COX10) causes cytochrome c oxidase deficiency. <i>Human Molecular Genetics</i> , 2000, 9, 1245-1249.	2.9	261
89	Expression of mutant alpha-synuclein causes increased susceptibility to dopamine toxicity. <i>Human Molecular Genetics</i> , 2000, 9, 2683-2689.	2.9	182
90	Altered gene expression and functions of mitochondria in human nephrotic syndrome. <i>FASEB Journal</i> , 1999, 13, 523-532.	0.5	53

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91	Decreased Brain Protein Levels of Cytochrome Oxidase Subunits in Alzheimer's Disease and in Hereditary Spinocerebella Ataxia Disorders. <i>Journal of Neurochemistry</i> , 1999, 72, 700-707.	3.9	76
92	Biochemical abnormalities and excitotoxicity in Huntington's disease brain. <i>Annals of Neurology</i> , 1999, 45, 25-32.	5.3	439
93	A Missense Mutation of Cytochrome Oxidase Subunit II Causes Defective Assembly and Myopathy. <i>American Journal of Human Genetics</i> , 1999, 65, 1030-1039.	6.2	131
94	Biochemical, genetic and immunoblot analyses of 17 patients with an isolated cytochrome c oxidase deficiency. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 1999, 1455, 35-44.	3.8	29
95	The mitochondrial genome: structure, transcription, translation and replication. <i>Biochimica Et Biophysica Acta - Bioenergetics</i> , 1999, 1410, 103-123.	1.0	1,220
96	Mitochondrial DNA Depletion Syndrome is Expressed in Amniotic Fluid Cell Cultures. <i>American Journal of Pathology</i> , 1999, 155, 67-70.	3.8	48
97	Mitochondrial DNA Mutations and Nuclear Mitochondrial Interactions in Human Disease. , 1999, , 635-663.		1
98	Assembly of cytochrome-c oxidase in cultured human cells. <i>FEBS Journal</i> , 1998, 254, 389-394.	0.2	211
99	Cytochrome c Oxidase subunit I microdeletion in a patient with motor neuron disease. <i>Annals of Neurology</i> , 1998, 43, 110-116.	5.3	251
100	Mitochondrial DNA transmission of the mitochondrial defect in Parkinson's disease. <i>Annals of Neurology</i> , 1998, 44, 177-186.	5.3	301
101	Liver failure associated with mitochondrial DNA depletion. <i>Journal of Hepatology</i> , 1998, 28, 556-563.	3.7	106
102	SCID mice containing muscle with human mitochondrial DNA mutations. An animal model for mitochondrial DNA defects.. <i>Journal of Clinical Investigation</i> , 1998, 102, 2090-2095.	8.2	9
103	Molecular Mechanisms in Mitochondrial DNA Depletion Syndrome. <i>Human Molecular Genetics</i> , 1997, 6, 935-942.	2.9	121
104	Expression of mtDNA and nDNA encoded respiratory chain proteins in chemically and genetically-derived Rho0 human fibroblasts: a comparison of subunit proteins in normal fibroblasts treated with ethidium bromide and fibroblasts from a patient with mtDNA depletion syndrome. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 1997, 1362, 145-159.	3.8	97
105	Human cytochrome c oxidase: structure, function, and deficiency. <i>Journal of Bioenergetics and Biomembranes</i> , 1997, 29, 151-163.	2.3	80
106	Subunit specific monoclonal antibodies show different steady-state levels of various cytochrome-c oxidase subunits in chronic progressive external ophthalmoplegia. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 1996, 1315, 199-207.	3.8	49
107	[9] Mammalian cytochrome-c oxidase: Characterization of enzyme and immunological detection of subunits in tissue extracts and whole cells. <i>Methods in Enzymology</i> , 1995, 260, 117-132.	1.0	121
108	Regulation of Cytochrome c Oxidase by Interaction of ATP at Two Binding Sites, One on Subunit VIa. <i>Biochemistry</i> , 1994, 33, 11833-11841.	2.5	54

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109	Regulation of the expression of mitochondrial proteins: relationship between mtDNA copy number and cytochrome-c oxidase activity in human cells and tissues. <i>Biochimica Et Biophysica Acta - Bioenergetics</i> , 1993, 1144, 177-183.	1.0	51
110	Steady-state transcript levels of cytochrome c oxidase genes during human myogenesis indicate subunit switching of subunit VIa and co-expression of subunit VIIa isoforms. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 1992, 1139, 155-162.	3.8	44
111	Identification of three human pseudogenes for subunit VIb of cytochrome c oxidase: a molecular record of gene evolution. <i>Gene</i> , 1991, 102, 237-244.	2.2	10
112	Nucleotide sequence of the last exon of the gene for human cytochrome c oxidase subunit VIb and its flanking regions. <i>Biochimica Et Biophysica Acta Gene Regulatory Mechanisms</i> , 1991, 1089, 283-285.	2.4	10
113	Assignment of the gene coding for human cytochrome c oxidase subunit VIb to chromosome 19, band q13.1, by fluorescence in situ hybridisation. <i>Human Genetics</i> , 1991, 87, 325-327.	3.8	18
114	Nucleotide sequence of cDNA encoding human fumarylacetoacetase. <i>Nucleic Acids Research</i> , 1990, 18, 1887-1887.	14.5	30
115	Isolation of cDNAs encoding subunit VIb of human cytochrome c oxidase and steady-state levels of coxVIb mRNA in different tissues. <i>Gene</i> , 1990, 93, 285-291.	2.2	19
116	Nucleotide sequence of cDNA encoding subunit VIb of human cytochromecoxidase. <i>Nucleic Acids Research</i> , 1989, 17, 1766-1766.	14.5	26
117	Type I Tyrosinemia: Lack of Immunologically Detectable Fumarylacetoacetase Enzyme Protein in Tissues and Cell Extracts. <i>Pediatric Research</i> , 1987, 22, 394-398.	2.3	34
118	Odour-conditioned anemotaxis of apterous aphids ( <i>Cryptomyzus korschelti</i> ) in response to host plants. <i>Physiological Entomology</i> , 1987, 12, 473-479.	1.5	36
119	Expression of Nodulin Genes During Nodule Development from Effective and Ineffective Root Nodules. , 1984, , 579-586.		9