

# Jan W Taanman

## List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

117 papers	7,518 citations	43 h-index	86 g-index
126 ext. papers	8,205 ext. citations	6.1 avg, IF	5.72 L-index

#	Paper	IF	Citations
117	Ambroxol reverses tau and Synuclein accumulation in a cholinergic N370S GBA1 mutation model.. <i>Human Molecular Genetics</i> , <b>2022</b> ,	5.6	2
116	The PINK1-Parkin mitophagy signalling pathway is not functional in peripheral blood mononuclear cells. <i>PLoS ONE</i> , <b>2021</b> , 16, e0259903	3.7	0
115	Mitochondrial respiratory chain and Krebs cycle enzyme function in human donor livers subjected to end-ischæmic hypothermic machine perfusion. <i>PLoS ONE</i> , <b>2021</b> , 16, e0257783	3.7	
114	Sirtuin 5 depletion impairs mitochondrial function in human proximal tubular epithelial cells. <i>Scientific Reports</i> , <b>2021</b> , 11, 15510	4.9	3
113	Mitochondria as target to inhibit proliferation and induce apoptosis of cancer cells: the effects of doxycycline and gemcitabine. <i>Scientific Reports</i> , <b>2020</b> , 10, 4363	4.9	31
112	Mitochondrial DNA: Structure, Genetics, Replication and Defects <b>2019</b> , 127-152		
111	Huntingtin Aggregates and Mitochondrial Pathology in Skeletal Muscle but not Heart of Late-Stage R6/2 Mice. <i>Journal of Huntington's Disease</i> , <b>2019</b> , 8, 145-159	1.9	9
110	NDUFA4 (Renamed COXFA4) Is a Cytochrome-c Oxidase Subunit. <i>Trends in Endocrinology and Metabolism</i> , <b>2018</b> , 29, 452-454	8.8	27
109	Somatic copy number gains of Synuclein (SNCA) in Parkinson's disease and multiple system atrophy brains. <i>Brain</i> , <b>2018</b> , 141, 2419-2431	11.2	41
108	Mitochondria as oncotarget: a comparison between the tetracycline analogs doxycycline and COL-3. <i>Oncotarget</i> , <b>2018</b> , 9, 33818-33831	3.3	13
107	A Human Neural Crest Stem Cell-Derived Dopaminergic Neuronal Model Recapitulates Biochemical Abnormalities in GBA1 Mutation Carriers. <i>Stem Cell Reports</i> , <b>2017</b> , 8, 728-742	8	42
106	Clinicopathologic and molecular spectrum of -related mitochondrial disease. <i>Neurology: Genetics</i> , <b>2017</b> , 3, e149	3.8	14
105	The clinical spectrum and natural history of early-onset diseases due to DNA polymerase gamma mutations. <i>Genetics in Medicine</i> , <b>2017</b> , 19, 1217-1225	8.1	32
104	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> , <b>2017</b> , 12, e0180467	3.7	18
103	Subunit composition of respiratory chain complex 1 and its responses to oxygen in mitochondria from human donor livers. <i>BMC Research Notes</i> , <b>2017</b> , 10, 547	2.3	1
102	Mitochondrial cristae remodelling is associated with disrupted OPA1 oligomerisation in the Huntington's disease R6/2 fragment model. <i>Experimental Neurology</i> , <b>2017</b> , 288, 167-175	5.7	17
101	Loss of PINK1 or Parkin Function Results in a Progressive Loss of Mitochondrial Function <b>2017</b> , 187-209		1

100	B25 Mitochondrial fission and fusion in skeletal muscle from HD patients and zQ175 mice. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2016</b> , 87, A17.3-A18	5.5	
99	A Clinical, Neuropathological and Genetic Study of Homozygous A467T POLG-Related Mitochondrial Disease. <i>PLoS ONE</i> , <b>2016</b> , 11, e0145500	3.7	33
98	A LON-ClpP Proteolytic Axis Degrades Complex I to Extinguish ROS Production in Depolarized Mitochondria. <i>Cell Reports</i> , <b>2016</b> , 17, 2522-2531	10.6	49
97	TRNT1 deficiency: clinical, biochemical and molecular genetic features. <i>Orphanet Journal of Rare Diseases</i> , <b>2016</b> , 11, 90	4.2	50
96	Selective striatal mtDNA depletion in end-stage Huntington's disease R6/2 mice. <i>Experimental Neurology</i> , <b>2015</b> , 266, 22-9	5.7	16
95	Clonal expansion of T cells in abdominal aortic aneurysm: a role for doxycycline as drug of choice?. <i>International Journal of Molecular Sciences</i> , <b>2015</b> , 16, 11178-95	6.3	12
94	Signal transducer and activator of transcription 2 deficiency is a novel disorder of mitochondrial fission. <i>Brain</i> , <b>2015</b> , 138, 2834-46	11.2	59
93	B34 Mitochondrial Biogenesis, And Respiratory Chain Assembly And Function, In Heart Of R6/2 Mouse Model Of Huntington's Disease. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2014</b> , 85, A21-A21	5.5	21
92	D03 Mitochondrial Biogenesis, And Respiratory Chain Assembly And Function, In Skeletal Muscle Of The R6/2 Mouse Model And Human Huntington's Disease. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2014</b> , 85, A32-A32	5.5	
91	Comment on "aneurysmal lesions of patients with abdominal aortic aneurysm contain clonally expanded T cells". <i>Journal of Immunology</i> , <b>2014</b> , 193, 2041	5.3	2
90	NDUFA4 mutations underlie dysfunction of a cytochrome c oxidase subunit linked to human neurological disease. <i>Cell Reports</i> , <b>2013</b> , 3, 1795-805	10.6	85
89	Pathogenic Parkinson's disease mutations across the functional domains of LRRK2 alter the autophagic/lysosomal response to starvation. <i>Biochemical and Biophysical Research Communications</i> , <b>2013</b> , 441, 862-6	3.4	66
88	HIBCH mutations can cause Leigh-like disease with combined deficiency of multiple mitochondrial respiratory chain enzymes and pyruvate dehydrogenase. <i>Orphanet Journal of Rare Diseases</i> , <b>2013</b> , 8, 188	4.2	57
87	NDUFA4 Mutations Underlie Dysfunction of a Cytochrome c Oxidase Subunit Linked to Human Neurological Disease. <i>Cell Reports</i> , <b>2013</b> , 4, 402	10.6	78
86	Glucosylceramidase degradation in fibroblasts carrying bi-allelic Parkin mutations. <i>Molecular Genetics and Metabolism</i> , <b>2013</b> , 109, 402-3	3.7	5
85	COX10 mutations resulting in complex multisystem mitochondrial disease that remains stable into adulthood. <i>JAMA Neurology</i> , <b>2013</b> , 70, 1556-61	17.2	24
84	Mitofusin 1 and mitofusin 2 are ubiquitinated in a PINK1/parkin-dependent manner upon induction of mitophagy. <i>Human Molecular Genetics</i> , <b>2013</b> , 22, 1697-1697	5.6	4
83	Creation of an open-access, mutation-defined fibroblast resource for neurological disease research. <i>PLoS ONE</i> , <b>2012</b> , 7, e43099	3.7	35

82	Pathogenic LRRK2 mutations do not alter gene expression in cell model systems or human brain tissue. <i>PLoS ONE</i> , <b>2011</b> , 6, e22489	3.7	27
81	Influence of zinc and zinc chelator on HT-29 colorectal cell line. <i>BioMetals</i> , <b>2011</b> , 24, 143-51	3.4	9
80	Kearns-Sayre syndrome caused by defective R1/p53R2 assembly. <i>Journal of Medical Genetics</i> , <b>2011</b> , 48, 610-7	5.8	41
79	Mutations in the mitochondrial complex I assembly factor NDUFAF1 cause fatal infantile hypertrophic cardiomyopathy. <i>Journal of Medical Genetics</i> , <b>2011</b> , 48, 691-7	5.8	55
78	14 Mitochondrial cardiomyopathy caused by defective assembly of respiratory chain complex I. <i>Heart</i> , <b>2011</b> , 97, e8-e8	5.1	
77	Parkinson's disease induced pluripotent stem cells with triplication of the $\beta$ -synuclein locus. <i>Nature Communications</i> , <b>2011</b> , 2, 440	17.4	328
76	Mitofusin 1 and mitofusin 2 are ubiquitinated in a PINK1/parkin-dependent manner upon induction of mitophagy. <i>Human Molecular Genetics</i> , <b>2010</b> , 19, 4861-70	5.6	680
75	FOXRED1, encoding an FAD-dependent oxidoreductase complex-I-specific molecular chaperone, is mutated in infantile-onset mitochondrial encephalopathy. <i>Human Molecular Genetics</i> , <b>2010</b> , 19, 4837-47	5.6	73
74	POG01 Anti-thymidine phosphorylase antibodies in the diagnosis of mitochondrial neurogastrointestinal encephalomyopathy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2010</b> , 81, e48-e48	5.5	
73	The diagnosis of inherited metabolic diseases by microarray gene expression profiling. <i>Orphanet Journal of Rare Diseases</i> , <b>2010</b> , 5, 34	4.2	2
72	Intracellular oxygenation and cytochrome oxidase C activity in ischemic preconditioning of steatotic rabbit liver. <i>American Journal of Surgery</i> , <b>2010</b> , 200, 507-18	2.7	7
71	Measurement of kinetic parameters of human platelet DNA polymerase gamma. <i>Methods</i> , <b>2010</b> , 51, 374-86	4.8	2
70	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> , <b>2010</b> , 1803, 931-9	4.9	50
69	Lowering the apoptotic threshold in colorectal cancer cells by targeting mitochondria. <i>Cancer Cell International</i> , <b>2010</b> , 10, 31	6.4	9
68	Analysis of mutant DNA polymerase gamma in patients with mitochondrial DNA depletion. <i>Human Mutation</i> , <b>2009</b> , 30, 248-54	4.7	46
67	Status epilepticus in children with Alpers' disease caused by POLG1 mutations: EEG and MRI features. <i>Epilepsia</i> , <b>2009</b> , 50, 1596-607	6.4	123
66	Increased sensitivity of myoblasts to oxidative stress in amyotrophic lateral sclerosis peripheral tissues. <i>Experimental Neurology</i> , <b>2009</b> , 218, 92-7	5.7	12
65	Differential effects of PINK1 nonsense and missense mutations on mitochondrial function and morphology. <i>Experimental Neurology</i> , <b>2009</b> , 219, 266-73	5.7	90

64	Collated mutations in mitochondrial DNA (mtDNA) depletion syndrome (excluding the mitochondrial gamma polymerase, POLG1). <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , <b>2009</b> , 1792, 1109-12	6.9	33
63	Characterization of a novel TYMP splice site mutation associated with mitochondrial neurogastrointestinal encephalomyopathy (MNGIE). <i>Neuromuscular Disorders</i> , <b>2009</b> , 19, 151-4	2.9	24
62	Does doxycycline work in synergy with cisplatin and oxaliplatin in colorectal cancer?. <i>World Journal of Surgical Oncology</i> , <b>2009</b> , 7, 2	3.4	14
61	Silencing of PINK1 expression affects mitochondrial DNA and oxidative phosphorylation in dopaminergic cells. <i>PLoS ONE</i> , <b>2009</b> , 4, e4756	3.7	147
60	Influence of mitochondrial DNA level on cellular energy metabolism: implications for mitochondrial diseases. <i>Journal of Bioenergetics and Biomembranes</i> , <b>2008</b> , 40, 59-67	3.7	48
59	Myoclonus-dystonia syndrome with severe depression is caused by an exon-skipping mutation in the epsilon-sarcoglycan gene. <i>Movement Disorders</i> , <b>2007</b> , 22, 1173-5	7	38
58	Relapsing neuropathy in an 18-year-old woman. <i>Lancet Neurology, The</i> , <b>2007</b> , 6, 192-8	24.1	4
57	Kinetic properties of mutant deoxyguanosine kinase in a case of reversible hepatic mtDNA depletion. <i>Biochemical Journal</i> , <b>2007</b> , 402, 377-85	3.8	34
56	Dominant inheritance of premature ovarian failure associated with mutant mitochondrial DNA polymerase gamma. <i>Human Reproduction</i> , <b>2006</b> , 21, 2467-73	5.7	132
55	Phenotypic variability of mitochondrial disease caused by a nuclear mutation in complex II. <i>Molecular Genetics and Metabolism</i> , <b>2006</b> , 89, 214-21	3.7	38
54	Assessment of the significance of mitochondrial DNA damage by chemotherapeutic agents <b>2005</b> , 27, 337		
53	Clinical, biochemical and morphological features of hepatocerebral syndrome with mitochondrial DNA depletion due to deoxyguanosine kinase deficiency. <i>Journal of Hepatology</i> , <b>2005</b> , 43, 333-41	13.4	69
52	Analysis of the trinucleotide CAG repeat from the DNA polymerase gamma gene (POLG) in patients with Parkinson's disease. <i>Neuroscience Letters</i> , <b>2005</b> , 376, 56-9	3.3	35
51	Analysis of COX2 mutants reveals cytochrome oxidase subassemblies in yeast. <i>Biochemical Journal</i> , <b>2005</b> , 390, 703-8	3.8	29
50	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> , <b>2005</b> , 20, 432-440	7	41
49	The Human Mitochondrial Genome. <i>Oxidative Stress and Disease</i> , <b>2005</b> , 95-246		1
48	Cytochrome c oxidase subassemblies in fibroblast cultures from patients carrying mutations in COX10, SCO1, or SURF1. <i>Journal of Biological Chemistry</i> , <b>2004</b> , 279, 7462-9	5.4	102
47	Replication of mitochondrial DNA occurs throughout the mitochondria of cultured human cells. <i>Experimental Cell Research</i> , <b>2003</b> , 289, 133-42	4.2	76

46	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> , <b>2003</b> , 12, 1839-45	5.6	71
45	A novel mutation in the deoxyguanosine kinase gene causing depletion of mitochondrial DNA. <i>Annals of Neurology</i> , <b>2002</b> , 52, 237-9	9.4	39
44	Diagnostic value of succinate ubiquinone reductase activity in the identification of patients with mitochondrial DNA depletion. <i>Journal of Inherited Metabolic Disease</i> , <b>2002</b> , 25, 7-16	5.4	23
43	Chapter 1 Structure and Function of the Mitochondrial Oxidative Phosphorylation System. <i>Blue Books of Practical Neurology</i> , <b>2002</b> , 1-34		1
42	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> , <b>2002</b> , 1554, 101-7	4.6	18
41	Depletion of mitochondrial DNA in the liver of an infant with neonatal giant cell hepatitis. <i>Human Pathology</i> , <b>2002</b> , 33, 247-53	3.7	42
40	Assembly of cytochrome c oxidase: what can we learn from patients with cytochrome c oxidase deficiency?. <i>Biochemical Society Transactions</i> , <b>2001</b> , 29, 446-51	5.1	29
39	Immunological phenotyping of fibroblast cultures from patients with a mitochondrial respiratory chain deficit. <i>Laboratory Investigation</i> , <b>2001</b> , 81, 1069-77	5.9	15
38	A mutant mitochondrial respiratory chain assembly protein causes complex III deficiency in patients with tubulopathy, encephalopathy and liver failure. <i>Nature Genetics</i> , <b>2001</b> , 29, 57-60	36.3	262
37	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> , <b>2001</b> , 73, 340-3	3.7	20
36	Mitochondrial dysfunction in congenital nephrotic syndrome. <i>Laboratory Investigation</i> , <b>2000</b> , 80, 1227-32	5.9	24
35	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> , <b>2000</b> , 9, 2733-42	5.6	93
34	A mutation in the human heme A:farnesyltransferase gene (COX10 ) causes cytochrome c oxidase deficiency. <i>Human Molecular Genetics</i> , <b>2000</b> , 9, 1245-9	5.6	223
33	Expression of mutant alpha-synuclein causes increased susceptibility to dopamine toxicity. <i>Human Molecular Genetics</i> , <b>2000</b> , 9, 2683-9	5.6	166
32	Altered gene expression and functions of mitochondria in human nephrotic syndrome. <i>FASEB Journal</i> , <b>1999</b> , 13, 523-32	0.9	47
31	Decreased brain protein levels of cytochrome oxidase subunits in Alzheimer's disease and in hereditary spinocerebellar ataxia disorders: a nonspecific change?. <i>Journal of Neurochemistry</i> , <b>1999</b> , 72, 700-7	6	62
30	Biochemical abnormalities and excitotoxicity in Huntington's disease brain. <i>Annals of Neurology</i> , <b>1999</b> , 45, 25-32	9.4	393
29	A missense mutation of cytochrome oxidase subunit II causes defective assembly and myopathy. <i>American Journal of Human Genetics</i> , <b>1999</b> , 65, 1030-9	11	120

28	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> , <b>1999</b> , 1455, 35-44	6.9	24
27	The mitochondrial genome: structure, transcription, translation and replication. <i>Biochimica Et Biophysica Acta - Bioenergetics</i> , <b>1999</b> , 1410, 103-23	4.6	940
26	Mitochondrial DNA depletion syndrome is expressed in amniotic fluid cell cultures. <i>American Journal of Pathology</i> , <b>1999</b> , 155, 67-70	5.8	45
25	Mitochondrial DNA Mutations and Nuclear Mitochondrial Interactions in Human Disease <b>1999</b> , 635-663		
24	Assembly of cytochrome-c oxidase in cultured human cells. <i>FEBS Journal</i> , <b>1998</b> , 254, 389-94		193
23	Cytochrome c oxidase subunit I microdeletion in a patient with motor neuron disease. <i>Annals of Neurology</i> , <b>1998</b> , 43, 110-6	9.4	226
22	Mitochondrial DNA transmission of the mitochondrial defect in Parkinson's disease. <i>Annals of Neurology</i> , <b>1998</b> , 44, 177-86	9.4	278
21	Liver failure associated with mitochondrial DNA depletion. <i>Journal of Hepatology</i> , <b>1998</b> , 28, 556-63	13.4	95
20	SCID mice containing muscle with human mitochondrial DNA mutations. An animal model for mitochondrial DNA defects. <i>Journal of Clinical Investigation</i> , <b>1998</b> , 102, 2090-5	15.9	8
19	Molecular mechanisms in mitochondrial DNA depletion syndrome. <i>Human Molecular Genetics</i> , <b>1997</b> , 6, 935-42	5.6	112
18	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> , <b>1997</b> , 1362, 145-59	6.9	86
17	Human cytochrome c oxidase: structure, function, and deficiency. <i>Journal of Bioenergetics and Biomembranes</i> , <b>1997</b> , 29, 151-63	3.7	68
16	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> , <b>1996</b> , 1315, 199-207	6.9	41
15	Mammalian cytochrome-c oxidase: characterization of enzyme and immunological detection of subunits in tissue extracts and whole cells. <i>Methods in Enzymology</i> , <b>1995</b> , 260, 117-32	1.7	109
14	Regulation of cytochrome c oxidase by interaction of ATP at two binding sites, one on subunit VIa. <i>Biochemistry</i> , <b>1994</b> , 33, 11833-41	3.2	51
13	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> , <b>1993</b> , 1144, 177-83	4.6	45
12	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> , <b>1992</b> , 1139, 155-62	6.9	39
11	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> , <b>1991</b> , 87, 325-7	6.3	17



10	Identification of three human pseudogenes for subunit VIb of cytochrome c oxidase: a molecular record of gene evolution. <i>Gene</i> , <b>1991</b> , 102, 237-44	3.8	7
9	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> , <b>1991</b> , 1089, 283-5		10
8	Nucleotide sequence of cDNA encoding human fumarylacetoacetase. <i>Nucleic Acids Research</i> , <b>1990</b> , 18, 1887	20.1	24
7	Isolation of cDNAs encoding subunit VIb of cytochrome c oxidase and steady-state levels of coxVIb mRNA in different tissues. <i>Gene</i> , <b>1990</b> , 93, 285-91	3.8	15
6	Nucleotide sequence of cDNA encoding subunit VIb of human cytochrome c oxidase. <i>Nucleic Acids Research</i> , <b>1989</b> , 17, 1766	20.1	22
5	Type I tyrosinemia: lack of immunologically detectable fumarylacetoacetase enzyme protein in tissues and cell extracts. <i>Pediatric Research</i> , <b>1987</b> , 22, 394-8	3.2	33
4	Odour-conditioned anemotaxis of apterous aphids ( <i>Cryptomyzus korschelti</i> ) in response to host plants. <i>Physiological Entomology</i> , <b>1987</b> , 12, 473-479	1.9	34
3	Expression of Nodulin Genes During Nodule Development from Effective and Ineffective Root Nodules <b>1984</b> , 579-586		8
2	The PINK1 /Parkin mitophagy signalling pathway is not functional in peripheral blood mononuclear cells		1
1	Cytochrome-c Oxidase (COX)		2