

# david thorburn

## List of Publications by Citations

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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.

91 papers	9,716 citations	53 h-index	92 g-index
92 ext. papers	10,983 ext. citations	9.6 avg, IF	5.93 L-index

#	Paper	IF	Citations
91	A mitochondrial protein compendium elucidates complex I disease biology. <i>Cell</i> , <b>2008</b> , 134, 112-23	56.2	1507
90	Molecular diagnosis of infantile mitochondrial disease with targeted next-generation sequencing. <i>Science Translational Medicine</i> , <b>2012</b> , 4, 118ra10	17.5	362
89	Minimum birth prevalence of mitochondrial respiratory chain disorders in children. <i>Brain</i> , <b>2003</b> , 126, 1905-12	11.2	339
88	Mitochondrial respiratory chain supercomplexes are destabilized in Barth Syndrome patients. <i>Journal of Molecular Biology</i> , <b>2006</b> , 361, 462-9	6.5	326
87	RAGE-induced cytosolic ROS promote mitochondrial superoxide generation in diabetes. <i>Journal of the American Society of Nephrology: JASN</i> , <b>2009</b> , 20, 742-52	12.7	323
86	High-throughput, pooled sequencing identifies mutations in NUBPL and FOXRED1 in human complex I deficiency. <i>Nature Genetics</i> , <b>2010</b> , 42, 851-8	36.3	292
85	Biochemical assays of respiratory chain complex activity. <i>Methods in Cell Biology</i> , <b>2007</b> , 80, 93-119	1.8	271
84	Accessory subunits are integral for assembly and function of human mitochondrial complex I. <i>Nature</i> , <b>2016</b> , 538, 123-126	50.4	260
83	Leigh syndrome: One disorder, more than 75 monogenic causes. <i>Annals of Neurology</i> , <b>2016</b> , 79, 190-203	9.4	259
82	Analysis of the assembly profiles for mitochondrial- and nuclear-DNA-encoded subunits into complex I. <i>Molecular and Cellular Biology</i> , <b>2007</b> , 27, 4228-37	4.8	202
81	The inheritance of mitochondrial DNA heteroplasmy: random drift, selection or both?. <i>Trends in Genetics</i> , <b>2000</b> , 16, 500-5	8.5	198
80	Mutation of the mitochondrial tyrosyl-tRNA synthetase gene, YARS2, causes myopathy, lactic acidosis, and sideroblastic anemia--MLASA syndrome. <i>American Journal of Human Genetics</i> , <b>2010</b> , 87, 52-9	11	190
79	Mitochondrial dysfunction in diabetic kidney disease. <i>Nature Reviews Nephrology</i> , <b>2018</b> , 14, 291-312	14.9	178
78	Human CIA30 is involved in the early assembly of mitochondrial complex I and mutations in its gene cause disease. <i>EMBO Journal</i> , <b>2007</b> , 26, 3227-37	13	172
77	Mitochondrial disorders: prevalence, myths and advances. <i>Journal of Inherited Metabolic Disease</i> , <b>2004</b> , 27, 349-62	5.4	171
76	Genetic counseling and prenatal diagnosis for the mitochondrial DNA mutations at nucleotide 8993. <i>American Journal of Human Genetics</i> , <b>1999</b> , 65, 474-82	11	165
75	Risk of developing a mitochondrial DNA deletion disorder. <i>Lancet, The</i> , <b>2004</b> , 364, 592-6	40	161

74	Skewed segregation of the mtDNA nt 8993 (T-->G) mutation in human oocytes. <i>American Journal of Human Genetics</i> , <b>1997</b> , 60, 1495-501	11	150
73	Mutation of C20orf7 disrupts complex I assembly and causes lethal neonatal mitochondrial disease. <i>American Journal of Human Genetics</i> , <b>2008</b> , 83, 468-78	11	150
72	De novo mutations in the mitochondrial ND3 gene as a cause of infantile mitochondrial encephalopathy and complex I deficiency. <i>Annals of Neurology</i> , <b>2004</b> , 55, 58-64	9.4	148
71	NDUFS6 mutations are a novel cause of lethal neonatal mitochondrial complex I deficiency. <i>Journal of Clinical Investigation</i> , <b>2004</b> , 114, 837-45	15.9	143
70	Mutations of the mitochondrial ND1 gene as a cause of MELAS. <i>Journal of Medical Genetics</i> , <b>2004</b> , 41, 784-9	5.8	135
69	Compensatory growth of healthy cardiac cells in the presence of diseased cells restores tissue homeostasis during heart development. <i>Developmental Cell</i> , <b>2008</b> , 15, 521-33	10.2	133
68	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
67	Mitochondrial energy generation disorders: genes, mechanisms, and clues to pathology. <i>Journal of Biological Chemistry</i> , <b>2019</b> , 294, 5386-5395	5.4	125
66	Mutations in MTFMT underlie a human disorder of formylation causing impaired mitochondrial translation. <i>Cell Metabolism</i> , <b>2011</b> , 14, 428-34	24.6	123
65	Monolysocardiolipins accumulate in Barth syndrome but do not lead to enhanced apoptosis. <i>Journal of Lipid Research</i> , <b>2005</b> , 46, 1182-95	6.3	108
64	Turn up the power - pharmacological activation of mitochondrial biogenesis in mouse models. <i>British Journal of Pharmacology</i> , <b>2014</b> , 171, 1818-36	8.6	89
63	IGF2BP2/IMP2-Deficient mice resist obesity through enhanced translation of Ucp1 mRNA and Other mRNAs encoding mitochondrial proteins. <i>Cell Metabolism</i> , <b>2015</b> , 21, 609-21	24.6	87
62	Analysis of mitochondrial subunit assembly into respiratory chain complexes using Blue Native polyacrylamide gel electrophoresis. <i>Analytical Biochemistry</i> , <b>2007</b> , 364, 128-37	3.1	86
61	Abundance of the POLG disease mutations in Europe, Australia, New Zealand, and the United States explained by single ancient European founders. <i>European Journal of Human Genetics</i> , <b>2007</b> , 15, 779-83	5.3	86
60	Biochemical and molecular diagnosis of mitochondrial respiratory chain disorders. <i>Biochimica Et Biophysica Acta - Bioenergetics</i> , <b>2004</b> , 1659, 121-8	4.6	85
59	Mitochondrial DNA mutations at nucleotide 8993 show a lack of tissue- or age-related variation. <i>Journal of Inherited Metabolic Disease</i> , <b>1999</b> , 22, 899-914	5.4	85
58	Mutations in LYRM4, encoding iron-sulfur cluster biogenesis factor ISD11, cause deficiency of multiple respiratory chain complexes. <i>Human Molecular Genetics</i> , <b>2013</b> , 22, 4460-73	5.6	81
57	Respiratory chain complex I deficiency caused by mitochondrial DNA mutations. <i>European Journal of Human Genetics</i> , <b>2011</b> , 19, 769-75	5.3	81

56	N-Acetylcysteine improves mitochondrial function and ameliorates behavioral deficits in the R6/1 mouse model of Huntington's disease. <i>Translational Psychiatry</i> , <b>2015</b> , 5, e492	8.6	80
55	Mutations in the UQCC1-interacting protein, UQCC2, cause human complex III deficiency associated with perturbed cytochrome b protein expression. <i>PLoS Genetics</i> , <b>2013</b> , 9, e1004034	6	79
54	ATAD3 gene cluster deletions cause cerebellar dysfunction associated with altered mitochondrial DNA and cholesterol metabolism. <i>Brain</i> , <b>2017</b> , 140, 1595-1610	11.2	76
53	Respiratory chain enzyme analysis in muscle and liver. <i>Mitochondrion</i> , <b>2004</b> , 4, 363-75	4.9	76
52	COA6 is a mitochondrial complex IV assembly factor critical for biogenesis of mtDNA-encoded COX2. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 5404-15	5.6	72
51	Fatal portal hypertension, liver failure, and mitochondrial dysfunction after HIV-1 nucleoside analogue-induced hepatitis and lactic acidemia. <i>Lancet, The</i> , <b>2001</b> , 357, 1412-4	4.0	72
50	A SLC39A8 variant causes manganese deficiency, and glycosylation and mitochondrial disorders. <i>Journal of Inherited Metabolic Disease</i> , <b>2017</b> , 40, 261-269	5.4	70
49	Mitochondrial myopathy with tRNA(Leu(UUR)) mutation and complex I deficiency responsive to riboflavin. <i>Journal of Pediatrics</i> , <b>1997</b> , 130, 138-45	3.6	69
48	Assembly of nuclear DNA-encoded subunits into mitochondrial complex IV, and their preferential integration into supercomplex forms in patient mitochondria. <i>FEBS Journal</i> , <b>2009</b> , 276, 6701-13	5.7	67
47	Mitochondrial oxidative phosphorylation disorders presenting in neonates: clinical manifestations and enzymatic and molecular diagnoses. <i>Pediatrics</i> , <b>2008</b> , 122, 1003-8	7.4	64
46	Mutations in the gene encoding 3-hydroxyisobutyryl-CoA hydrolase results in progressive infantile neurodegeneration. <i>American Journal of Human Genetics</i> , <b>2007</b> , 80, 195-9	11	64
45	Regulation of the human-erythrocyte hexose-monophosphate shunt under conditions of oxidative stress. A study using NMR spectroscopy, a kinetic isotope effect, a reconstituted system and computer simulation. <i>FEBS Journal</i> , <b>1985</b> , 150, 371-86		62
44	Cardiac manifestations in oxidative phosphorylation disorders of childhood. <i>Journal of Pediatrics</i> , <b>2007</b> , 150, 407-11	3.6	60
43	Biallelic Mutations in MRPS34 Lead to Instability of the Small Mitochondrial Subunit and Leigh Syndrome. <i>American Journal of Human Genetics</i> , <b>2017</b> , 101, 239-254	11	59
42	Evidence for the toxicity of bidirectional transcripts and mitochondrial dysfunction in blood associated with small CGG expansions in the FMR1 gene in patients with parkinsonism. <i>Genetics in Medicine</i> , <b>2011</b> , 13, 392-9	8.1	59
41	The p.M292T NDUFS2 mutation causes complex I-deficient Leigh syndrome in multiple families. <i>Brain</i> , <b>2010</b> , 133, 2952-63	11.2	58
40	Mitochondrial Disease Sequence Data Resource (MSeqDR): a global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities. <i>Molecular Genetics and Metabolism</i> , <b>2015</b> , 114, 388-96	3.7	56
39	Characterization of mitochondrial FOXRED1 in the assembly of respiratory chain complex I. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 2952-65	5.6	54

38	Beneficial effects of resveratrol on respiratory chain defects in patients. Fibroblasts involve estrogen receptor and estrogen-related receptor alpha signaling. <i>Human Molecular Genetics</i> , <b>2014</b> , 23, 2106-19	5.6	52
37	MPV17 Loss Causes Deoxynucleotide Insufficiency and Slow DNA Replication in Mitochondria. <i>PLoS Genetics</i> , <b>2016</b> , 12, e1005779	6	52
36	Mutations in the gene encoding C8orf38 block complex I assembly by inhibiting production of the mitochondria-encoded subunit ND1. <i>Journal of Molecular Biology</i> , <b>2011</b> , 414, 413-26	6.5	50
35	Approaches to finding the molecular basis of mitochondrial oxidative phosphorylation disorders. <i>Twin Research and Human Genetics</i> , <b>2008</b> , 11, 395-411	2.2	50
34	A founder mutation in PET100 causes isolated complex IV deficiency in Lebanese individuals with Leigh syndrome. <i>American Journal of Human Genetics</i> , <b>2014</b> , 94, 209-22	11	49
33	Mutation in mitochondrial ribosomal protein S7 (MRPS7) causes congenital sensorineural deafness, progressive hepatic and renal failure and lactic acidemia. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 2297-307	5.6	48
32	Mutations in CYC1, encoding cytochrome c1 subunit of respiratory chain complex III, cause insulin-responsive hyperglycemia. <i>American Journal of Human Genetics</i> , <b>2013</b> , 93, 384-9	11	48
31	Next-generation sequencing in molecular diagnosis: NUBPL mutations highlight the challenges of variant detection and interpretation. <i>Human Mutation</i> , <b>2012</b> , 33, 411-8	4.7	47
30	Comparison of computer simulations of the F-type and L-type non-oxidative hexose monophosphate shunts with <sup>31</sup> P-NMR experimental data from human erythrocytes. <i>FEBS Journal</i> , <b>1989</b> , 180, 399-420		47
29	The molecular basis of malonyl-CoA decarboxylase deficiency. <i>American Journal of Human Genetics</i> , <b>1999</b> , 65, 318-26	11	45
28	Biallelic Mutations in TMEM126B Cause Severe Complex I Deficiency with a Variable Clinical Phenotype. <i>American Journal of Human Genetics</i> , <b>2016</b> , 99, 217-27	11	45
27	Mutation screening of the mitochondrial genome using denaturing high-performance liquid chromatography. <i>Molecular Genetics and Metabolism</i> , <b>2005</b> , 84, 61-74	3.7	44
26	Late-adult onset Leigh syndrome. <i>Journal of Clinical Neuroscience</i> , <b>2012</b> , 19, 195-202	2.2	42
25	Proteomic and metabolomic analyses of mitochondrial complex I-deficient mouse model generated by spontaneous B2 short interspersed nuclear element (SINE) insertion into NADH dehydrogenase (ubiquinone) Fe-S protein 4 (Ndufs4) gene. <i>Journal of Biological Chemistry</i> , <b>2012</b> , 287, 20652-63	5.4	42
24	Recent advances in the genetics of mitochondrial encephalopathies. <i>Current Neurology and Neuroscience Reports</i> , <b>2010</b> , 10, 277-85	6.6	42
23	Human BAC-mediated rescue of the Friedreich ataxia knockout mutation in transgenic mice. <i>Mammalian Genome</i> , <b>2004</b> , 15, 370-82	3.2	41
22	Serine protease inhibition and mitochondrial dysfunction associated with cisplatin resistance in human tumor cell lines: targets for therapy. <i>Biochemical Pharmacology</i> , <b>1997</b> , 53, 1673-82	6	36
21	HIV-1 protein Vpr causes gross mitochondrial dysfunction in the yeast <i>Saccharomyces cerevisiae</i> . <i>FEBS Letters</i> , <b>1997</b> , 410, 145-9	3.8	36

20	Monolysocardiolipin in cultured fibroblasts is a sensitive and specific marker for Barth Syndrome. <i>Journal of Lipid Research</i> , <b>2006</b> , 47, 2346-51	6.3	35
19	Deficiency in Apoptosis-Inducing Factor Recapitulates Chronic Kidney Disease via Aberrant Mitochondrial Homeostasis. <i>Diabetes</i> , <b>2016</b> , 65, 1085-98	0.9	34
18	Decreased activities of mitochondrial respiratory chain complexes in non-mitochondrial respiratory chain diseases. <i>Developmental Medicine and Child Neurology</i> , <b>2006</b> , 48, 132-6	3.3	34
17	Juvenile Alpers disease. <i>Archives of Neurology</i> , <b>2008</b> , 65, 121-4		33
16	Biparental inheritance of mitochondrial DNA in humans is not a common phenomenon. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 2823-2826	8.1	23
15	Fatal perinatal mitochondrial cardiac failure caused by recurrent duplications in the locus. <i>Med</i> , <b>2021</b> , 2, 49-73	31.7	15
14	Early diagnosis of Pearson syndrome in neonatal intensive care following rapid mitochondrial genome sequencing in tandem with exome sequencing. <i>European Journal of Human Genetics</i> , <b>2019</b> , 27, 1821-1826	5.3	12
13	Loss of BIM increases mitochondrial oxygen consumption and lipid oxidation, reduces adiposity and improves insulin sensitivity in mice. <i>Cell Death and Differentiation</i> , <b>2018</b> , 25, 217-225	12.7	11
12	Application of oligonucleotide array CGH in the detection of a large intragenic deletion in POLG associated with Alpers Syndrome. <i>Mitochondrion</i> , <b>2011</b> , 11, 104-7	4.9	11
11	Leigh syndrome caused by mutations in is associated with a better prognosis. <i>Annals of Clinical and Translational Neurology</i> , <b>2019</b> , 6, 515-524	5.3	9
10	Multitomic analysis elucidates Complex I deficiency caused by a deep intronic variant in NDUFB10. <i>Human Mutation</i> , <b>2021</b> , 42, 19-24	4.7	9
9	A patient with homozygous nonsense variants in two Leigh syndrome disease genes: Distinguishing a dual diagnosis from a hypomorphic protein-truncating variant. <i>Human Mutation</i> , <b>2019</b> , 40, 893-898	4.7	7
8	High-intensity training induces non-stoichiometric changes in the mitochondrial proteome of human skeletal muscle without reorganisation of respiratory chain content. <i>Nature Communications</i> , <b>2021</b> , 12, 7056	17.4	7
7	Modelling Mitochondrial Disease in Human Pluripotent Stem Cells: What Have We Learned?. <i>International Journal of Molecular Sciences</i> , <b>2021</b> , 22,	6.3	5
6	Application of Genome Sequencing from Blood to Diagnose Mitochondrial Diseases. <i>Genes</i> , <b>2021</b> , 12,	4.2	3
5	A novel variant in COX16 causes cytochrome c oxidase deficiency, severe fatal neonatal lactic acidosis, encephalopathy, cardiomyopathy, and liver dysfunction. <i>Human Mutation</i> , <b>2021</b> , 42, 135-141	4.7	2
4	Sideroflexin 4 is a complex I assembly factor that interacts with the MCIA complex and is required for the assembly of the ND2 module.. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2022</b> , 119, e2115566119	11.5	2
3	Biallelic Variants in Cause a Severe Infantile Metabolic Disorder Affecting Mitochondrial Function.. <i>International Journal of Molecular Sciences</i> , <b>2022</b> , 23,	6.3	1

- 2 Genomic sequencing for the diagnosis of childhood mitochondrial disorders: a health economic evaluation. *European Journal of Human Genetics*, **2021**, 5:3 1
- 1 Distinct diagnostic trajectories in NBAS-associated acute liver failure highlights the need for timely functional studies.. *JIMD Reports*, **2022**, 63, 240-249 1:9