david thorburn

List of Publications by Year in Descending Order

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	Biallelic Variants in Cause a Severe Infantile Metabolic Disorder Affecting Mitochondrial Function.. *International Journal of Molecular Sciences*, **2022**, 23,	6.3	1
90	Sideroflexin 4 is a complex I assembly factor that interacts with the MCIA complex and is required for the assembly of the ND2 module.. *Proceedings of the National Academy of Sciences of the United States of America*, **2022**, 119, e2115566119	11.5	2
89	Distinct diagnostic trajectories in NBAS-associated acute liver failure highlights the need for timely functional studies.. *JIMD Reports*, **2022**, 63, 240-249	1.9	
88	High-intensity training induces non-stoichiometric changes in the mitochondrial proteome of human skeletal muscle without reorganisation of respiratory chain content. *Nature Communications* , **2021**, 12, 7056	17.4	7
87	Application of Genome Sequencing from Blood to Diagnose Mitochondrial Diseases. *Genes*, **2021**, 12,	4.2	3
86	Genomic sequencing for the diagnosis of childhood mitochondrial disorders: a health economic evaluation. *European Journal of Human Genetics*, **2021**,	5.3	1
85	Modelling Mitochondrial Disease in Human Pluripotent Stem Cells: What Have We Learned?. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	5
84	Multiomic analysis elucidates Complex I deficiency caused by a deep intronic variant in NDUFB10. *Human Mutation*, **2021**, 42, 19-24	4.7	9
83	Fatal perinatal mitochondrial cardiac failure caused by recurrent duplications in the locus. *Med*, **2021**, 2, 49-73	31.7	15
82	A novel variant in COX16 causes cytochrome c oxidase deficiency, severe fatal neonatal lactic acidosis, encephalopathy, cardiomyopathy, and liver dysfunction. *Human Mutation*, **2021**, 42, 135-141	4.7	2
81	Biparental inheritance of mitochondrial DNA in humans is not a common phenomenon. *Genetics in Medicine*, **2019**, 21, 2823-2826	8.1	23
80	Leigh syndrome caused by mutations in is associated with a better prognosis. *Annals of Clinical and Translational Neurology*, **2019**, 6, 515-524	5.3	9
79	A patient with homozygous nonsense variants in two Leigh syndrome disease genes: Distinguishing a dual diagnosis from a hypomorphic protein-truncating variant. *Human Mutation*, **2019**, 40, 893-898	4.7	7
78	Early diagnosis of Pearson syndrome in neonatal intensive care following rapid mitochondrial genome sequencing in tandem with exome sequencing. *European Journal of Human Genetics*, **2019**, 27, 1821-1826	5.3	12
77	Mitochondrial energy generation disorders: genes, mechanisms, and clues to pathology. *Journal of Biological Chemistry*, **2019**, 294, 5386-5395	5.4	125
76	Mitochondrial dysfunction in diabetic kidney disease. *Nature Reviews Nephrology*, **2018**, 14, 291-312	14.9	178
75	Loss of BIM increases mitochondrial oxygen consumption and lipid oxidation, reduces adiposity and improves insulin sensitivity in mice. *Cell Death and Differentiation*, **2018**, 25, 217-225	12.7	11

74	A SLC39A8 variant causes manganese deficiency, and glycosylation and mitochondrial disorders. *Journal of Inherited Metabolic Disease*, **2017**, 40, 261-269	5.4	70
73	ATAD3 gene cluster deletions cause cerebellar dysfunction associated with altered mitochondrial DNA and cholesterol metabolism. *Brain*, **2017**, 140, 1595-1610	11.2	76
72	Biallelic Mutations in MRPS34 Lead to Instability of the Small Mitoribosomal Subunit and Leigh Syndrome. *American Journal of Human Genetics*, **2017**, 101, 239-254	11	59
71	Accessory subunits are integral for assembly and function of human mitochondrial complex I. *Nature*, **2016**, 538, 123-126	50.4	260
70	Deficiency in Apoptosis-Inducing Factor Recapitulates Chronic Kidney Disease via Aberrant Mitochondrial Homeostasis. *Diabetes*, **2016**, 65, 1085-98	0.9	34
69	MPV17 Loss Causes Deoxynucleotide Insufficiency and Slow DNA Replication in Mitochondria. *PLoS Genetics*, **2016**, 12, e1005779	6	52
68	Biallelic Mutations in TMEM126B Cause Severe Complex I Deficiency with a Variable Clinical Phenotype. *American Journal of Human Genetics*, **2016**, 99, 217-27	11	45
67	Leigh syndrome: One disorder, more than 75 monogenic causes. *Annals of Neurology*, **2016**, 79, 190-203	9.4	259
66	Mutation in mitochondrial ribosomal protein S7 (MRPS7) causes congenital sensorineural deafness, progressive hepatic and renal failure and lactic acidemia. *Human Molecular Genetics*, **2015**, 24, 2297-307	5.6	48
65	COA6 is a mitochondrial complex IV assembly factor critical for biogenesis of mtDNA-encoded COX2. *Human Molecular Genetics*, **2015**, 24, 5404-15	5.6	72
64	IGF2BP2/IMP2-Deficient mice resist obesity through enhanced translation of Ucp1 mRNA and Other mRNAs encoding mitochondrial proteins. *Cell Metabolism*, **2015**, 21, 609-21	24.6	87
63	N-Acetylcysteine improves mitochondrial function and ameliorates behavioral deficits in the R6/1 mouse model of Huntington's disease. *Translational Psychiatry*, **2015**, 5, e492	8.6	80
62	Characterization of mitochondrial FOXRED1 in the assembly of respiratory chain complex I. *Human Molecular Genetics*, **2015**, 24, 2952-65	5.6	54
61	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. *Molecular Genetics and Metabolism*, **2015**, 114, 388-96	3.7	56
60	Turn up the power - pharmacological activation of mitochondrial biogenesis in mouse models. *British Journal of Pharmacology*, **2014**, 171, 1818-36	8.6	89
59	A founder mutation in PET100 causes isolated complex IV deficiency in Lebanese individuals with Leigh syndrome. *American Journal of Human Genetics*, **2014**, 94, 209-22	11	49
58	Beneficial effects of resveratrol on respiratory chain defects in patients' fibroblasts involve estrogen receptor and estrogen-related receptor alpha signaling. *Human Molecular Genetics*, **2014**, 23, 2106-19	5.6	52
57	Mutations in CYC1, encoding cytochrome c1 subunit of respiratory chain complex III, cause insulin-responsive hyperglycemia. *American Journal of Human Genetics*, **2013**, 93, 384-9	11	48

56	Mutations in LYRM4, encoding iron-sulfur cluster biogenesis factor ISD11, cause deficiency of multiple respiratory chain complexes. *Human Molecular Genetics*, **2013**, 22, 4460-73	5.6	81
55	Mutations in the UQCC1-interacting protein, UQCC2, cause human complex III deficiency associated with perturbed cytochrome b protein expression. *PLoS Genetics*, **2013**, 9, e1004034	6	79
54	Next-generation sequencing in molecular diagnosis: NUBPL mutations highlight the challenges of variant detection and interpretation. *Human Mutation*, **2012**, 33, 411-8	4.7	47
53	Molecular diagnosis of infantile mitochondrial disease with targeted next-generation sequencing. *Science Translational Medicine*, **2012**, 4, 118ra10	17.5	362
52	Late-adult onset Leigh syndrome. *Journal of Clinical Neuroscience*, **2012**, 19, 195-202	2.2	42
51	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. *Journal of Biological Chemistry*, **2012**, 287, 20652-63	5.4	42
50	Mutations in MTFMT underlie a human disorder of formylation causing impaired mitochondrial translation. *Cell Metabolism*, **2011**, 14, 428-34	24.6	123
49	Mutations in the gene encoding C8orf38 block complex I assembly by inhibiting production of the mitochondria-encoded subunit ND1. *Journal of Molecular Biology*, **2011**, 414, 413-26	6.5	50
48	Respiratory chain complex I deficiency caused by mitochondrial DNA mutations. *European Journal of Human Genetics*, **2011**, 19, 769-75	5.3	81
47	Application of oligonucleotide array CGH in the detection of a large intragenic deletion in POLG associated with Alpers Syndrome. *Mitochondrion*, **2011**, 11, 104-7	4.9	11
46	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. *Genetics in Medicine*, **2011**, 13, 392-9	8.1	59
45	High-throughput, pooled sequencing identifies mutations in NUBPL and FOXRED1 in human complex I deficiency. *Nature Genetics*, **2010**, 42, 851-8	36.3	292
44	The p.M292T NDUFS2 mutation causes complex I-deficient Leigh syndrome in multiple families. *Brain*, **2010**, 133, 2952-63	11.2	58
43	Mutation of the mitochondrial tyrosyl-tRNA synthetase gene, YARS2, causes myopathy, lactic acidosis, and sideroblastic anemia--MLASA syndrome. *American Journal of Human Genetics*, **2010**, 87, 52-9	11	190
42	Recent advances in the genetics of mitochondrial encephalopathies. *Current Neurology and Neuroscience Reports*, **2010**, 10, 277-85	6.6	42
41	RAGE-induced cytosolic ROS promote mitochondrial superoxide generation in diabetes. *Journal of the American Society of Nephrology: JASN*, **2009**, 20, 742-52	12.7	323
40	Assembly of nuclear DNA-encoded subunits into mitochondrial complex IV, and their preferential integration into supercomplex forms in patient mitochondria. *FEBS Journal*, **2009**, 276, 6701-13	5.7	67
39	A mitochondrial protein compendium elucidates complex I disease biology. *Cell*, **2008**, 134, 112-23	56.2	1507

38	Compensatory growth of healthy cardiac cells in the presence of diseased cells restores tissue homeostasis during heart development. *Developmental Cell*, **2008**, 15, 521-33	10.2	133
37	Approaches to finding the molecular basis of mitochondrial oxidative phosphorylation disorders. *Twin Research and Human Genetics*, **2008**, 11, 395-411	2.2	50
36	Mitochondrial oxidative phosphorylation disorders presenting in neonates: clinical manifestations and enzymatic and molecular diagnoses. *Pediatrics*, **2008**, 122, 1003-8	7.4	64
35	Juvenile Alpers disease. *Archives of Neurology*, **2008**, 65, 121-4		33
34	Mutation of C20orf7 disrupts complex I assembly and causes lethal neonatal mitochondrial disease. *American Journal of Human Genetics*, **2008**, 83, 468-78	11	150
33	Biochemical assays of respiratory chain complex activity. *Methods in Cell Biology*, **2007**, 80, 93-119	1.8	271
32	Analysis of mitochondrial subunit assembly into respiratory chain complexes using Blue Native polyacrylamide gel electrophoresis. *Analytical Biochemistry*, **2007**, 364, 128-37	3.1	86
31	Abundance of the POLG disease mutations in Europe, Australia, New Zealand, and the United States explained by single ancient European founders. *European Journal of Human Genetics*, **2007**, 15, 779-83	5.3	86
30	Human CIA30 is involved in the early assembly of mitochondrial complex I and mutations in its gene cause disease. *EMBO Journal*, **2007**, 26, 3227-37	13	172
29	Cardiac manifestations in oxidative phosphorylation disorders of childhood. *Journal of Pediatrics*, **2007**, 150, 407-11	3.6	60
28	Analysis of the assembly profiles for mitochondrial- and nuclear-DNA-encoded subunits into complex I. *Molecular and Cellular Biology*, **2007**, 27, 4228-37	4.8	202
27	Mutations in the gene encoding 3-hydroxyisobutyryl-CoA hydrolase results in progressive infantile neurodegeneration. *American Journal of Human Genetics*, **2007**, 80, 195-9	11	64
26	Monolysocardiolipin in cultured fibroblasts is a sensitive and specific marker for Barth Syndrome. *Journal of Lipid Research*, **2006**, 47, 2346-51	6.3	35
25	Dominant inheritance of premature ovarian failure associated with mutant mitochondrial DNA polymerase gamma. *Human Reproduction*, **2006**, 21, 2467-73	5.7	132
24	Mitochondrial respiratory chain supercomplexes are destabilized in Barth Syndrome patients. *Journal of Molecular Biology*, **2006**, 361, 462-9	6.5	326
23	Decreased activities of mitochondrial respiratory chain complexes in non-mitochondrial respiratory chain diseases. *Developmental Medicine and Child Neurology*, **2006**, 48, 132-6	3.3	34
22	Mutation screening of the mitochondrial genome using denaturing high-performance liquid chromatography. *Molecular Genetics and Metabolism*, **2005**, 84, 61-74	3.7	44
21	Monolysocardiolipins accumulate in Barth syndrome but do not lead to enhanced apoptosis. *Journal of Lipid Research*, **2005**, 46, 1182-95	6.3	108

20	Mitochondrial disorders: prevalence, myths and advances. *Journal of Inherited Metabolic Disease*, **2004**, 27, 349-62	5.4	171
19	Human BAC-mediated rescue of the Friedreich ataxia knockout mutation in transgenic mice. *Mammalian Genome*, **2004**, 15, 370-82	3.2	41
18	De novo mutations in the mitochondrial ND3 gene as a cause of infantile mitochondrial encephalopathy and complex I deficiency. *Annals of Neurology*, **2004**, 55, 58-64	9.4	148
17	Mutations of the mitochondrial ND1 gene as a cause of MELAS. *Journal of Medical Genetics*, **2004**, 41, 784-9	5.8	135
16	Biochemical and molecular diagnosis of mitochondrial respiratory chain disorders. *Biochimica Et Biophysica Acta - Bioenergetics*, **2004**, 1659, 121-8	4.6	85
15	Respiratory chain enzyme analysis in muscle and liver. *Mitochondrion*, **2004**, 4, 363-75	4.9	76
14	Risk of developing a mitochondrial DNA deletion disorder. *Lancet, The*, **2004**, 364, 592-6	40	161
13	NDUFS6 mutations are a novel cause of lethal neonatal mitochondrial complex I deficiency. *Journal of Clinical Investigation*, **2004**, 114, 837-45	15.9	143
12	Minimum birth prevalence of mitochondrial respiratory chain disorders in children. *Brain*, **2003**, 126, 1905-12	11	339
11	Fatal portal hypertension, liver failure, and mitochondrial dysfunction after HIV-1 nucleoside analogue-induced hepatitis and lactic acidaemia. *Lancet, The*, **2001**, 357, 1412-4	40	72
10	The inheritance of mitochondrial DNA heteroplasmy: random drift, selection or both?. *Trends in Genetics*, **2000**, 16, 500-5	8.5	198
9	Mitochondrial DNA mutations at nucleotide 8993 show a lack of tissue- or age-related variation. *Journal of Inherited Metabolic Disease*, **1999**, 22, 899-914	5.4	85
8	Genetic counseling and prenatal diagnosis for the mitochondrial DNA mutations at nucleotide 8993. *American Journal of Human Genetics*, **1999**, 65, 474-82	11	165
7	The molecular basis of malonyl-CoA decarboxylase deficiency. *American Journal of Human Genetics*, **1999**, 65, 318-26	11	45
6	Serine protease inhibition and mitochondrial dysfunction associated with cisplatin resistance in human tumor cell lines: targets for therapy. *Biochemical Pharmacology*, **1997**, 53, 1673-82	6	36
5	Mitochondrial myopathy with tRNA(Leu(UUR)) mutation and complex I deficiency responsive to riboflavin. *Journal of Pediatrics*, **1997**, 130, 138-45	3.6	69
4	HIV-1 protein Vpr causes gross mitochondrial dysfunction in the yeast Saccharomyces cerevisiae. *FEBS Letters*, **1997**, 410, 145-9	3.8	36
3	Skewed segregation of the mtDNA nt 8993 (T-->G) mutation in human oocytes. *American Journal of Human Genetics*, **1997**, 60, 1495-501	11	150

2	Comparison of computer simulations of the F-type and L-type non-oxidative hexose monophosphate shunts with 31P-NMR experimental data from human erythrocytes. *FEBS Journal*, **1989**, 180, 399-420	47
1	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. *FEBS Journal*, **1985**, 150, 371-86	62