Ann Saada

List of Publications by Year
in descending order

172 papers	9,741 citations	31974 53 h-index	42393 92 g-index
185 all docs	185 docs citations	185 times ranked	11820 citing authors

#	Article	IF	Citations
1	Mutant mitochondrial thymidine kinase in mitochondrial DNA depletion myopathy. Nature Genetics, 2001, 29, 342-344.	21.4	551
2	The deoxyguanosine kinase gene is mutated in individuals with depleted hepatocerebral mitochondrial DNA. Nature Genetics, 2001, 29, 337-341.	21.4	521
3	Deleterious Mutation in the Mitochondrial Arginylâ€“Transfer RNA Synthetase Gene Is Associated with Pontocerebellar Hypoplasia. American Journal of Human Genetics, 2007, 81, 857-862.	6.2	306
4	Deficiency of the ADP-Forming Succinyl-CoA Synthase Activity Is Associated with Encephalomyopathy and Mitochondrial DNA Depletion. American Journal of Human Genetics, 2005, 76, 1081-1086.	6.2	284
5	Control of Pancreatic Î² Cell Regeneration by Glucose Metabolism. Cell Metabolism, 2011, 13, 440-449.	16.2	266
6	Peripheral nerve injury induces Schwann cells to express two macrophage phenotypes: phagocytosis and the galactose-specific lectin MAC-2. Journal of Neuroscience, 1994, 14, 3231-3245.	3.6	208
7	Defective mitochondrial translation caused by a ribosomal protein (MRPS16) mutation. Annals of Neurology, 2004, 56, 734-738.	5.3	205
8	Acute Infantile Liver Failure Due to Mutations in the TRMU Gene. American Journal of Human Genetics, 2009, 85, 401-407.	6.2	205
9	Mutations in the Fatty Acid 2-Hydroxylase Gene Are Associated with Leukodystrophy with Spastic Paraparesis and Dystonia. American Journal of Human Genetics, 2008, 83, 643-648.	6.2	193
10	Mutations in LPIN1 Cause Recurrent Acute Myoglobinuria in Childhood. American Journal of Human Genetics, 2008, 83, 489-494.	6.2	189
11	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
12	Ablation of Ceramide Synthase 2 Causes Chronic Oxidative Stress Due to Disruption of the Mitochondrial Respiratory Chain. Journal of Biological Chemistry, 2013, 288, 4947-4956.	3.4	165
13	Large-scale implementation of pooled RNA extraction and RT-PCR for SARS-CoV-2 detection. Clinical Microbiology and Infection, 2020, 26, 1248-1253.	6.0	164
14	C6ORF66 Is an Assembly Factor of Mitochondrial Complex I. American Journal of Human Genetics, 2008, 82, 32-38.	6.2	155
15	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
16	An international classification of inherited metabolic disorders (<scp>ICIMD</scp>). Journal of Inherited Metabolic Disease, 2021, 44, 164-177.	3.6	146
17	Antenatal mitochondrial disease caused by mitochondrial ribosomal protein (MRPS22) mutation. Journal of Medical Genetics, 2007, 44, 784-786.	3.2	144
18	Mutations in GTPBP3 Cause a Mitochondrial Translation Defect Associated with Hypertrophic Cardiomyopathy, Lactic Acidosis, and Encephalopathy. American Journal of Human Genetics, 2014, 95, 708-720.	6.2	123

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19	Type 2 Diabetes and Congenital Hyperinsulinism Cause DNA Double-Strand Breaks and p53 Activity in Î² Cells. Cell Metabolism, 2014, 19, 109-121.	16.2	123
20	Weaning Triggers a Maturation Step of Pancreatic Î² Cells. Developmental Cell, 2015, 32, 535-545.	7.0	120
21	Ceramide and the mitochondrial respiratory chain. Biochimie, 2014, 100, 88-94.	2.6	117
22	Mitochondrial Complex III Deficiency Associated with a Homozygous Mutation in UQCRQ. American Journal of Human Genetics, 2008, 82, 1211-1216.	6.2	114
23	FASTKD2 Nonsense Mutation in an Infantile Mitochondrial Encephalomyopathy Associated with Cytochrome C Oxidase Deficiency. American Journal of Human Genetics, 2008, 83, 415-423.	6.2	107
24	Mitochondrial complex I deficiency caused by a deleterious NDUFA11 mutation. Annals of Neurology, 2008, 63, 405-408.	5.3	103
25	Apoptosis-Like Death, an Extreme SOS Response in Escherichia coli. MBio, 2014, 5, e01426-14.	4.1	102
26	Granulocyte macrophage colony stimulating factor produced in lesioned peripheral nerves induces the up-regulation of cell surface expression of MAC-2 by macrophages and Schwann cells.. Journal of Cell Biology, 1996, 133, 159-167.	5.2	100
27	Familial neonatal isolated cardiomyopathy caused by a mutation in the flavoprotein subunit of succinate dehydrogenase. European Journal of Human Genetics, 2010, 18, 1160-1165.	2.8	100
28	The Transgenic Overexpression of Î±-Synuclein and Not Its Related Pathology Associates with Complex I Inhibition. Journal of Biological Chemistry, 2010, 285, 7334-7343.	3.4	96
29	Screening for Active Small Molecules in Mitochondrial Complex I Deficient Patient's Fibroblasts, Reveals AICAR as the Most Beneficial Compound. PLoS ONE, 2011, 6, e26883.	2.5	95
30	Infantile Cerebellar-Retinal Degeneration Associated with a Mutation in Mitochondrial Aconitase, ACO2. American Journal of Human Genetics, 2012, 90, 518-523.	6.2	93
31	Recurrent De Novo Dominant Mutations in SLC25A4 Cause Severe Early-Onset Mitochondrial Disease and Loss of Mitochondrial DNA Copy Number. American Journal of Human Genetics, 2016, 99, 860-876.	6.2	93
32	Mice Deficient in Ribosomal Protein S6 Phosphorylation Suffer from Muscle Weakness that Reflects a Growth Defect and Energy Deficit. PLoS ONE, 2009, 4, e5618.	2.5	92
33	Fatal infantile mitochondrial encephalomyopathy, hypertrophic cardiomyopathy and optic atrophy associated with a homozygousOPA1mutation. Journal of Medical Genetics, 2016, 53, 127-131.	3.2	91
34	Mutation in mitochondrial ribosomal protein MRPS22 leads to Cornelia de Lange-like phenotype, brain abnormalities and hypertrophic cardiomyopathy. European Journal of Human Genetics, 2011, 19, 394-399.	2.8	90
35	Characteristics of Mitochondrial Transformation into Human Cells. Scientific Reports, 2016, 6, 26057.	3.3	90
36	mtDNA depletion myopathy: elucidation of the tissue specificity in the mitochondrial thymidine kinase (TK2) deficiency. Molecular Genetics and Metabolism, 2003, 79, 1-5.	1.1	89

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37	Early prenatal ventriculomegaly due to an AIFM1 mutation identified by linkage analysis and whole exome sequencing. Molecular Genetics and Metabolism, 2011, 104, 517-520.	1.1	89
38	Molecular basis of lipoamide dehydrogenase deficiency in Ashkenazi Jews. , 1999, 82, 177-182.		87
39	Mitochondrial complex IV deficiency, caused by mutated COX6B1, is associated with encephalomyopathy, hydrocephalus and cardiomyopathy. European Journal of Human Genetics, 2015, 23, 159-164.	2.8	82
40	Postnatal microcephaly and pain insensitivity due to a de novo heterozygous DNM1L mutation causing impaired mitochondrial fission and function. American Journal of Medical Genetics, Part A, 2016, 170, 1603-1607.	1.2	80
41	Exocrine Pancreatic Insufficiency, Dyserythropoeitic Anemia, and Calvarial Hyperostosis Are Caused by a Mutation in the COX4I2 Gene. American Journal of Human Genetics, 2009, 84, 412-417.	6.2	78
42	Mitochondrial respiratory enzymes are a major target of iron toxicity in rat heart cells. Translational Research, 1998, 131, 466-474.	2.3	74
43	The cytokine network of Wallerian degeneration: ILâ€10 and GMâ€CSF. European Journal of Neuroscience, 1998, 10, 2707-2713.	2.6	73
44	The Thr224Asn mutation in the VPS45 gene is associated with the congenital neutropenia and primary myelofibrosis of infancy. Blood, 2013, 121, 5078-5087.	1.4	70
45	Mitochondrial Transfer Ameliorates Cognitive Deficits, Neuronal Loss, and Gliosis in Alzheimerâ€™s Disease Mice. Journal of Alzheimer's Disease, 2019, 72, 587-604.	2.6	70
46	Kinetic Properties of Mutant Human Thymidine Kinase 2 Suggest a Mechanism for Mitochondrial DNA Depletion Myopathy. Journal of Biological Chemistry, 2003, 278, 6963-6968.	3.4	69
47	Deleterious mutation in FDX1L gene is associated with a novel mitochondrial muscle myopathy. European Journal of Human Genetics, 2014, 22, 902-906.	2.8	65
48	The effect of mutated mitochondrial ribosomal proteins S16 and S22 on the assembly of the small and large ribosomal subunits in human mitochondria. Mitochondrion, 2008, 8, 254-261.	3.4	62
49	Mitochondrial STAT3 plays a major role in IgE-antigenâ€“mediated mast cell exocytosis. Journal of Allergy and Clinical Immunology, 2014, 134, 460-469.e10.	2.9	62
50	Revisiting mitochondrial diagnostic criteria in the new era of genomics. Genetics in Medicine, 2018, 20, 444-451.	2.4	62
51	TMEM70 mutations are a common cause of nuclear encoded ATP synthase assembly defect: further delineation of a new syndrome. Journal of Medical Genetics, 2011, 48, 177-182.	3.2	61
52	Agenesis of corpus callosum and optic nerve hypoplasia due to mutations inSLC25A1encoding the mitochondrial citrate transporter. Journal of Medical Genetics, 2013, 50, 240-245.	3.2	60
53	A novel thiol antioxidant that crosses the blood brain barrier protects dopaminergic neurons in experimental models of Parkinson's disease. European Journal of Neuroscience, 2005, 21, 637-646.	2.6	59
54	Evaluation of enzymatic assays and compounds affecting ATP production in mitochondrial respiratory chain complex I deficiency. Analytical Biochemistry, 2004, 335, 66-72.	2.4	56

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55	Depletion of the other genome-mitochondrial DNA depletion syndromes in humans. Journal of Molecular Medicine, 2002, 80, 389-396.	3.9	54
56	NDUFS4 mutations cause Leigh syndrome with predominant brainstem involvement. Molecular Genetics and Metabolism, 2009, 97, 185-189.	1.1	54
57	Combined OXPHOS complex I and IV defect, due to mutated complex I assembly factor C20ORF7. Journal of Inherited Metabolic Disease, 2012, 35, 125-131.	3.6	52
58	Lipoamide Dehydrogenase Deficiency Due to a Novel Mutation in the Interface Domain. Biochemical and Biophysical Research Communications, 1999, 262, 163-166.	2.1	51
59	Antenatal presentation of carnitine palmitoyltransferase II deficiency. American Journal of Medical Genetics Part A, 2001, 102, 183-187.	2.4	50
60	Evaluating the therapeutic potential of idebenone and related quinone analogues in Leber hereditary optic neuropathy. Mitochondrion, 2017, 36, 36-42.	3.4	50
61	The interplay between SUCLA2, SUCLG2, and mitochondrial DNA depletion. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2011, 1812, 625-629.	3.8	49
62	Cap-independent translation by DAP5 controls cell fate decisions in human embryonic stem cells. Genes and Development, 2016, 30, 1991-2004.	5.9	49
63	Delineation of C12orf65-related phenotypes: a genotypeâ€“phenotype relationship. European Journal of Human Genetics, 2014, 22, 1019-1025.	2.8	48
64	A homozygous founder missense variant in arylsulfatase G abolishes its enzymatic activity causing atypical Usher syndrome in humans. Genetics in Medicine, 2018, 20, 1004-1012.	2.4	48
65	Cytogenetic analyses of premature ovarian failure using karyotyping and interphase fluorescence in situ hybridization (FISH) in a group of 1000 patients. Clinical Genetics, 2010, 78, 181-185.	2.0	47
66	The unique neuroradiology of complex I deficiency due to NDUFA12L defect. Molecular Genetics and Metabolism, 2008, 94, 78-82.	1.1	46
67	Coenzyme Qâ€“dependent mitochondrial respiratory chain activity in granulosa cells is reduced with aging. Fertility and Sterility, 2015, 104, 724-727.	1.0	45
68	The Effect of Antiepileptic Drugs on Mitochondrial Activity: A Pilot Study. Journal of Child Neurology, 2010, 25, 541-545.	1.4	44
69	l-arginine:glycine amidinotransferase (AGAT) deficiency: Clinical presentation and response to treatment in two patients with a novel mutation. Molecular Genetics and Metabolism, 2010, 101, 228-232.	1.1	44
70	Leukoencephalopathy with accumulated succinate is indicative of SDHAF1 related complex II deficiency. Orphanet Journal of Rare Diseases, 2012, 7, 69.	2.7	44
71	Mitochondrial hepato-encephalopathy due to deficiency of QIL1/MIC13 (C19orf70), a MICOS complex subunit. European Journal of Human Genetics, 2016, 24, 1778-1782.	2.8	44
72	Pathogenic variants in glutamyl-tRNAGln amidotransferase subunits cause a lethal mitochondrial cardiomyopathy disorder. Nature Communications, 2018, 9, 4065.	12.8	44

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73	Mitochondrial deoxyribonucleoside triphosphate pools in thymidine kinase 2 deficiency. Biochemical and Biophysical Research Communications, 2003, 310, 963-966.	2.1	41
74	Deoxyribonucleotides and Disorders of Mitochondrial DNA Integrity. DNA and Cell Biology, 2004, 23, 797-806.	1.9	41
75	Mutated NDUFS6 is the cause of fatal neonatal lactic acidemia in Caucasus Jews. European Journal of Human Genetics, 2009, 17, 1200-1203.	2.8	41
76	Diagnostic serum glycosylation profile in patients with intellectual disability as a result of MAN1B1 deficiency. Brain, 2014, 137, 1030-1038.	7.6	41
77	Hypomyelination and developmental delay associated withVPS11mutation in Ashkenazi-Jewish patients. Journal of Medical Genetics, 2015, 52, 749-753.	3.2	41
78	Infantile mitochondrial hepatopathy is a cardinal feature of MEGDEL syndrome (3â€Methylglutaconic) Tj ETQq0 0 0 rgBT /Overlock 10 Tf mutations in SERAC1. American Journal of Medical Genetics, Part A, 2013, 161, 2204-2215.	1.2	39
79	TAT-mediated Delivery of LAD Restores Pyruvate Dehydrogenase Complex Activity in the Mitochondria of Patients with LAD Deficiency. Molecular Therapy, 2008, 16, 691-697.	8.2	38
80	Mitochondria: Mitochondrial OXPHOS (dys) function ex vivo â€“ The use of primary fibroblasts. International Journal of Biochemistry and Cell Biology, 2014, 48, 60-65.	2.8	38
81	Mutation in the COX4I1 gene is associated with short stature, poor weight gain and increased chromosomal breaks, simulating Fanconi anemia. European Journal of Human Genetics, 2017, 25, 1142-1146.	2.8	38
82	The Effects of Ascorbate, N-Acetylcysteine, and Resveratrol on Fibroblasts from Patients with Mitochondrial Disorders. Journal of Clinical Medicine, 2017, 6, 1.	2.4	38
83	Antibiotic effects on mitochondrial translation and in patients with mitochondrial translational defects. Mitochondrion, 2009, 9, 429-437.	3.4	37
84	Mitochondrial Encephalomyopathy Associated with a Novel Mutation in the Mitochondrial tRNAleu(UUR)Gene (A3243T). Biochemical and Biophysical Research Communications, 1997, 233, 637-639.	2.1	36
85	Î±â€Synuclein abnormalities in mouse models of peroxisome biogenesis disorders. Journal of Neuroscience Research, 2010, 88, 866-876.	2.9	36
86	Protection of a Ceramide Synthase 2 Null Mouse from Drug-induced Liver Injury. Journal of Biological Chemistry, 2013, 288, 30904-30916.	3.4	35
87	Mitochondrial deoxyribonucleotide pools in deoxyguanosine kinase deficiency. Molecular Genetics and Metabolism, 2008, 95, 169-173.	1.1	33
88	Fulminant neurological deterioration in a neonate with Leigh syndrome due to a maternally transmitted missense mutation in the mitochondrial ND3 gene. Biochemical and Biophysical Research Communications, 2005, 334, 582-587.	2.1	32
89	Upregulation of Mitochondrial Content in Cytochrome c Oxidase Deficient Fibroblasts. PLoS ONE, 2016, 11, e0165417.	2.5	29
90	Effect of various agents on adenosine triphosphate synthesis in mitochondrial complex I deficiency. Journal of Pediatrics, 2001, 139, 868-870.	1.8	28

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91	Mitochondrial epileptic encephalopathy, 3â€methylglutaconic aciduria and variable complex V deficiency associated with <scp>TIMM50</scp> mutations. Clinical Genetics, 2017, 91, 690-696.	2.0	28
92	Development of pheochromocytoma in ceramide synthase 2 null mice. Endocrine-Related Cancer, 2015, 22, 623-632.	3.1	27
93	Lesion-Induced Changes in the Production of Newly Synthesized and Secreted Apo-E and Other Molecules Are Independent of the Concomitant Recruitment of Blood-Borne Macrophages into Injured Peripheral Nerves. Journal of Neurochemistry, 1992, 59, 1287-1292.	3.9	26
94	Two transgenic mouse models for Î²-subunit components of succinate-CoA ligase yielding pleiotropic metabolic alterations. Biochemical Journal, 2016, 473, 3463-3485.	3.7	26
95	Lipoamide dehydrogenase deficiency in Ashkenazi Jews: An insertion mutation in the mitochondrial leader sequence. Human Mutation, 1997, 10, 256-257.	2.5	25
96	The use of individual patient's fibroblasts in the search for personalized treatment of nuclear encoded OXPHOS diseases. Molecular Genetics and Metabolism, 2011, 104, 39-47.	1.1	24
97	Rat Cardiac Mitochondrial Sub-populations Show Distinct Features of Oxidative Phosphorylation during Ischemia, Reperfusion and Ischemic Preconditioning. Cellular Physiology and Biochemistry, 2012, 30, 83-94.	1.6	24
98	Nonylphenol Ethoxylate Plastic Additives Inhibit Mitochondrial Respiratory Chain Complex I. Clinical Chemistry, 2009, 55, 1883-1884.	3.2	23
99	2-hydroxylated sphingomyelin profiles in cells from patients with mutated fatty acid 2-hydroxylase. Lipids in Health and Disease, 2011, 10, 84.	3.0	23
100	Mitochondrial performance in heat acclimationâ€"a lesson from ischemia/reperfusion and calcium overload insults in the heart. American Journal of Physiology - Regulatory Integrative and Comparative Physiology, 2012, 303, R870-R881.	1.8	23
101	Dietary copper supplementation restores Î²-cell function of Cohen diabetic rats: a link between mitochondrial function and glucose-stimulated insulin secretion. American Journal of Physiology - Endocrinology and Metabolism, 2013, 304, E1023-E1034.	3.5	23
102	The effect of small molecules on nuclear-encoded translation diseases. Biochimie, 2014, 100, 184-191.	2.6	23
103	Adherence of Ureaplasma urealyticum to human erythrocytes. Infection and Immunity, 1991, 59, 467-469.	2.2	23
104	ATP Synthesis in Lipoamide Dehydrogenase Deficiency. Biochemical and Biophysical Research Communications, 2000, 269, 382-386.	2.1	22
105	Leigh disease presenting in utero due to a novel missense mutation in the mitochondrial DNAâ€"ND3. Molecular Genetics and Metabolism, 2010, 100, 65-70.	1.1	22
106	Human granulosa luteal cell oxidative phosphorylation function is not affected by age or ovarian response. Fertility and Sterility, 2012, 98, 166-172.e2.	1.0	22
107	Replacement of the C6ORF66 Assembly Factor (NDUFAF4) Restores Complex I Activity in Patient Cells. Molecular Medicine, 2013, 19, 124-134.	4.4	22
108	Nemaline Rods and Complex I Deficiency in Three Infants with Hypotonia, Motor Delay and Failure to Thrive. Neuropediatrics, 2004, 35, 302-306.	0.6	21

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109	Mitochondrial dysfunction in preclinical genetic prion disease: A target for preventive treatment?. Neurobiology of Disease, 2019, 124, 57-66.	4.4	21
110	Toward genotype phenotype correlations in GFM1 mutations. Mitochondrion, 2012, 12, 242-247.	3.4	20
111	Bezafibrate Improves Mitochondrial Fission and Function in DNM1L-Deficient Patient Cells. Cells, 2020, 9, 301.	4.1	20
112	Liver Disease in the Ashkenazi-Jewish Lipoamide Dehydrogenase Deficiency. Journal of Pediatric Gastroenterology and Nutrition, 1997, 24, 599-601.	1.8	19
113	Cardiac-Targeted Transgenic Mutant Mitochondrial Enzymes: mtDNA Defects, Antiretroviral Toxicity and Cardiomyopathy. Cardiovascular Toxicology, 2008, 8, 57-69.	2.7	17
114	Lipoamide dehydrogenase activity in lymphocytes. Clinica Chimica Acta, 1996, 256, 197-201.	1.1	16
115	Deoxyribonucleoside Kinases in Mitochondrial DNA Depletion. Nucleosides, Nucleotides and Nucleic Acids, 2004, 23, 1205-1215.	1.1	16
116	Is the aging human ovary still ticking?: Expression of clock-genes in luteinized granulosa cells of young and older women. Journal of Ovarian Research, 2018, 11, 95.	3.0	16
117	Calmodulin Methyltransferase Is Required for Growth, Muscle Strength, Somatosensory Development and Brain Function. PLoS Genetics, 2015, 11, e1005388.	3.5	16
118	Fibroblasts that Reside in Mouse and Frog Injured Peripheral Nerves Produce Apolipoproteins. Journal of Neurochemistry, 2002, 64, 1996-2003.	3.9	15
119	Homozygous p.V116* mutation inC12orf65results in Leigh syndrome. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, jnnp-2014-310084.	1.9	15
120	Novel Homozygous Missense Mutation in SPG20 Gene Results in Troyer Syndrome Associated with Mitochondrial Cytochrome c Oxidase Deficiency. JIMD Reports, 2016, 33, 55-60.	1.5	15
121	Oxidative stress elicited by modifying the ceramide acyl chain length reduces the rate of clathrin-mediated endocytosis. Journal of Cell Science, 2017, 130, 1486-1493.	2.0	15
122	Pathological presentation of cardiac mitochondria in a rat model for chronic kidney disease. PLoS ONE, 2018, 13, e0198196.	2.5	15
123	Primary Coenzyme Q deficiency Due to Novel ADCK3 Variants, Studies in Fibroblasts and Review of Literature. Neurochemical Research, 2019, 44, 2372-2384.	3.3	15
124	Elevated plasma citrulline: look for dihydrolipoamide dehydrogenase deficiency. European Journal of Pediatrics, 2014, 173, 243-245.	2.7	14
125	Cytochrome c oxidase deficiency, oxidative stress, possible antioxidant therapy and link to nuclear DNA damage. European Journal of Human Genetics, 2018, 26, 579-581.	2.8	14
126	A novel homozygous SLC25A1 mutation with impaired mitochondrial complex V: Possible phenotypic expansion. American Journal of Medical Genetics, Part A, 2018, 176, 330-336.	1.2	14

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127	A novel TUFM homozygous variant in a child with mitochondrial cardiomyopathy expands the phenotype of combined oxidative phosphorylation deficiency 4. Journal of Human Genetics, 2019, 64, 589-595.	2.3	14
128	Clinical Characteristics and Muscle Pathology in Myopathic Mitochondrial DNA Depletion. Journal of Child Neurology, 2002, 17, 499-504.	1.4	13
129	Glycosidase Activities of Mycoplasmas. Zentralblatt Fur Bakteriologie: International Journal of Medical Microbiology, 1990, 273, 300-305.	0.5	12
130	IL-1Î² hampers glucose-stimulated insulin secretion in Cohen diabetic rat islets through mitochondrial cytochromecoxidase inhibition by nitric oxide. American Journal of Physiology - Endocrinology and Metabolism, 2014, 306, E648-E657.	3.5	12
131	The Relationship between Mitochondrial Respiratory Chain Activities in Muscle and Metabolites in Plasma and Urine: A Retrospective Study. Journal of Clinical Medicine, 2017, 6, 31.	2.4	12
132	The pathomechanism of cytochrome c oxidase deficiency includes nuclear DNA damage. Biochimica Et Biophysica Acta - Bioenergetics, 2018, 1859, 893-900.	1.0	12
133	De novo pathogenic DNM1L variant in a patient diagnosed with atypical hereditary sensory and autonomic neuropathy. Molecular Genetics & Genomic Medicine, 2019, 7, e00961.	1.2	12
134	Fishing in the (deoxyribonucleotide) pool. Biochemical Journal, 2009, 422, e3-e6.	3.7	11
135	Infantile SOD1 deficiency syndrome caused by a homozygous SOD1 variant with absence of enzyme activity. Brain, 2022, 145, 872-878.	7.6	10
136	Mitochondrial OXPHOS function is unaffected by chronic azithromycin treatment. Journal of Cystic Fibrosis, 2013, 12, 682-687.	0.7	9
137	Upregulation of COX4-2 via HIF-1Î± in Mitochondrial COX4-1 Deficiency. Cells, 2021, 10, 452.	4.1	9
138	Novel selective human mitochondrial kinase inhibitors: Design, synthesis and enzymatic activity. Bioorganic and Medicinal Chemistry, 2007, 15, 3065-3081.	3.0	8
139	Heat acclimation mediated cardioprotection is controlled by mitochondrial metabolic remodeling involving HIF-1Î±. Journal of Thermal Biology, 2020, 93, 102691.	2.5	8
140	PF-4708671 Activates AMPK Independently of p70S6K1 Inhibition. PLoS ONE, 2014, 9, e107364.	2.5	8
141	Opposing effects of intracellular vs. extracellular adenine nucleotides on autophagy: implications for Î²-cell function. Journal of Cell Science, 2018, 131, .	2.0	7
142	Cytochrome c Oxidase Activity as a Metabolic Regulator in Pancreatic Beta-Cells. Cells, 2022, 11, 929.	4.1	7
143	Reversible fulminant lactic acidosis and liver failure in an infant with hepatic cytochrome-c oxidase deficiency. Journal of Inherited Metabolic Disease, 2002, 25, 371-377.	3.6	6
144	Severe infantile type of carnitine palmitoyltransferase II (CPT II) deficiency due to homozygous R503C mutation. Journal of Inherited Metabolic Disease, 2007, 30, 266-266.	3.6	6

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145	Biochemical assays of TCA cycle and Î²-oxidation metabolites. Methods in Cell Biology, 2020, 155, 83-120.	1.1	6
146	The role of orotic acid measurement in routine newborn screening for urea cycle disorders. Journal of Inherited Metabolic Disease, 2021, 44, 606-617.	3.6	6
147	Sea squirt alternative oxidase bypasses fatal mitochondrial heart disease. EMBO Molecular Medicine, 2019, 11, .	6.9	5
148	Treatment of ErbB2 breast cancer by mitochondrial targeting. Cancer & Metabolism, 2020, 8, 17.	5.0	5
149	A recurring NFS1 pathogenic variant causes a mitochondrial disorder with variable intra-familial patient outcomes. Molecular Genetics and Metabolism Reports, 2021, 26, 100699.	1.1	5
150	Levodopa-responsive dystonia caused by biallelic PRKN exon inversion invisible to exome sequencing. Brain Communications, 2021, 3, fcab197.	3.3	5
151	Insights into deoxyribonucleoside therapy for mitochondrial TK2 deficient mtDNA depletion. EBioMedicine, 2019, 47, 14-15.	6.1	4
152	Severe infantile epileptic encephalopathy associated with D-glyceric aciduria: report of a novel case and review. Metabolic Brain Disease, 2019, 34, 557-563.	2.9	4
153	A novel de novo heterozygous pathogenic variant in the SDHA gene results in childhood onset bilateral optic atrophy and cognitive impairment. Metabolic Brain Disease, 2021, 36, 581-588.	2.9	4
154	Replicative Stress Coincides with Impaired Nuclear DNA Damage Response in COX4-1 Deficiency. International Journal of Molecular Sciences, 2022, 23, 4149.	4.1	4
155	A novel homozygous MSTO1 mutation in Ashkenazi Jewish siblings with ataxia and myopathy. Journal of Human Genetics, 2021, 66, 835-840.	2.3	3
156	Comparing antiâ€“aging hallmark activities of Metformin and Nano-PSO in a mouse model of genetic Creutzfeldt-Jakob Disease. Neurobiology of Aging, 2022, 110, 77-87.	3.1	3
157	Interaction of a monoclonal antibody with the urease ofUreaplasma urealyticum. FEMS Microbiology Letters, 1988, 55, 187-190.	1.8	2
158	Weaning Triggers a Maturation Step of Pancreatic Î² Cells. Developmental Cell, 2015, 33, 238-239.	7.0	2
159	Multifaceted Analyses of Isolated Mitochondria Establish the Anticancer Drug 2-Hydroxyoleic Acid as an Inhibitor of Substrate Oxidation and an Activator of Complex IV-Dependent State 3 Respiration. Cells, 2022, 11, 578.	4.1	2
160	Mutations in LPIN1 Cause Recurrent Acute Myoglobinuria in Childhood. American Journal of Human Genetics, 2009, 84, 95.	6.2	1
161	The influence of in vivo exposure to nonylphenol ethoxylate 10 (NP-10) on the ovarian reserve in a mouse model. Reproductive Toxicology, 2018, 81, 246-252.	2.9	1
162	Multiple Acyl-CoA Dehydrogenase Deficiency with Variable Presentation Due to a Homozygous Mutation in a Bedouin Tribe. Genes, 2021, 12, 1140.	2.4	1

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163	Heat acclimation improves mitochondrial function following ischemia reperfusion insult. FASEB Journal, 2009, 23, 793.7.	0.5	1
164	Acute Infantile Liver Failure Due to Mutations in the TRMU Gene. American Journal of Human Genetics, 2010, 86, 295.	6.2	0
165	Complex II Deficiency. , 2016, , 265-272.		0
166	TAT-mediated Delivery of LAD Restores Pyruvate Dehydrogenase Complex Activity in the Mitochondria of Patients with LAD Deficiency. Molecular Therapy, 0, , .	8.2	0
167	Complex Subunits and Assembly Genes: Complex I. , 2013, , 185-202.		0
168	Abstract 381: Prolonged Renal Failure Leads to Reduced Number of Active Cardiac Mitochondria in a Rat Model for Long Term Chronic Kidney Disease. Circulation Research, 2015, 117, .	4.5	0
169	Measurement of troponin-T in dried blood spots and dried plasma spots: A pilot study. Journal of Pediatric Biochemistry, 2015, 04, 153-157.	0.2	0
170	Quantitative measurement of urinary glycosaminoglycans using a modified DMB method facilitates the diagnosis and monitoring of mucopolysaccharidoses. Journal of Pediatric Biochemistry, 2015, 02, 163-167.	0.2	0
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