Massimo Zeviani

List of Publications by Year
in descending order

491 papers	46,207 citations	1301 109 h-index	2684 193 g-index
518 all docs	518 docs citations	518 times ranked	29590 citing authors

#	Article	IF	Citations
1	Mitochondrial Neurodegeneration. Cells, 2022, 11, 637.	4.1	29
2	The relevance of migraine in the clinical spectrum of mitochondrial disorders. Scientific Reports, 2022, 12, 4222.	3.3	7
3	Mitochondrial Retinopathies. International Journal of Molecular Sciences, 2022, 23, 210.	4.1	29
4	Mitochondrial Cytochrome c Oxidase Defects Alter Cellular Homeostasis of Transition Metals. Frontiers in Cell and Developmental Biology, 2022, 10, .	3.7	5
5	Loss of function of the mitochondrial peptidase PITRM1 induces proteotoxic stress and Alzheimerâ€™s disease-like pathology in human cerebral organoids. Molecular Psychiatry, 2021, 26, 5733-5750.	7.9	79
6	Mitochondrial disorders of the OXPHOS system. FEBS Letters, 2021, 595, 1062-1106.	2.8	117
7	Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE): Position paper on diagnosis, prognosis, and treatment by the <scp>MNGIE</scp> International Network. Journal of Inherited Metabolic Disease, 2021, 44, 376-387.	3.6	47
8	Cytochrome c oxidase deficiency. Biochimica Et Biophysica Acta - Bioenergetics, 2021, 1862, 148335.	1.0	53
9	An international classification of inherited metabolic disorders (<scp>ICIMD</scp>). Journal of Inherited Metabolic Disease, 2021, 44, 164-177.	3.6	146
10	Loss of COX4I1 Leads to Combined Respiratory Chain Deficiency and Impaired Mitochondrial Protein Synthesis. Cells, 2021, 10, 369.	4.1	21
11	NDUFS3 depletion permits complex I maturation and reveals TMEM126A/OPA7 as an assembly factor binding the ND4-module intermediate. Cell Reports, 2021, 35, 109002.	6.4	13
12	Exploiting pyocyanin to treat mitochondrial disease due to respiratory complex III dysfunction. Nature Communications, 2021, 12, 2103.	12.8	16
13	Neural stem cells traffic functional mitochondria via extracellular vesicles. PLoS Biology, 2021, 19, e3001166.	5.6	95
14	DNA polymerase gamma mutations that impair holoenzyme stability cause catalytic subunit depletion. Nucleic Acids Research, 2021, 49, 5230-5248.	14.5	15
15	Myopathic mitochondrial DNA depletion syndrome associated with biallelic variants in LIG3. Brain, 2021, 144, e74-e74.	7.6	5
16	Duplexing complexome profiling with SILAC to study human respiratory chain assembly defects. Biochimica Et Biophysica Acta - Bioenergetics, 2021, 1862, 148395.	1.0	15
17	SILAC-based complexome profiling dissects the structural organization of the human respiratory supercomplexes in SCAFIKO cells. Biochimica Et Biophysica Acta - Bioenergetics, 2021, 1862, 148414.	1.0	15
18	Modelling of BCS1L-related human mitochondrial disease in Drosophila melanogaster. Journal of Molecular Medicine, 2021, 99, 1471-1485.	3.9	7

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19	Role of PITRM1 in Mitochondrial Dysfunction and Neurodegeneration. Biomedicines, 2021, 9, 833.	3.2	17
20	A de novo mutation in mitochondrial ATPsynthase subunit Î± causes a life threatening disease in neonates which heals in infancy. European Journal of Human Genetics, 2021, 29, 1593-1594.	2.8	0
21	Mitochondrial Structure and Bioenergetics in Normal and Disease Conditions. International Journal of Molecular Sciences, 2021, 22, 586.	4.1	72
22	Blue-Native Electrophoresis to Study the OXPHOS Complexes. Methods in Molecular Biology, 2021, 2192, 287-311.	0.9	17
23	Mutation in the MICOS subunit gene APOO (MIC26) associated with an X-linked recessive mitochondrial myopathy, lactic acidosis, cognitive impairment and autistic features. Journal of Medical Genetics, 2021, 58, 155-167.	3.2	28
24	Bi-allelic variants in OGDHL cause a neurodevelopmental spectrum disease featuring epilepsy, hearing loss, visual impairment, and ataxia. American Journal of Human Genetics, 2021, 108, 2368-2384.	6.2	12
25	Respiratory supercomplexes act as a platform for complex <scp>III</scp> â€mediated maturation of human mitochondrial complexes I and <scp>IV</scp>. EMBO Journal, 2020, 39, e102817.	7.8	102
26	Novel compound heterozygous pathogenic variants in nucleotide-binding protein like protein (NUBPL) cause leukoencephalopathy with multi-systemic involvement. Molecular Genetics and Metabolism, 2020, 129, 26-34.	1.1	9
27	Neurodevelopmental regression, severe generalized dystonia, and metabolic acidosis caused by POLR3A mutations. Neurology: Genetics, 2020, 6, e521.	1.9	4
28	Ethylmalonic encephalopathy: Clinical course and therapy response in an uncommon mild case with a severe ETHE1 mutation. Molecular Genetics and Metabolism Reports, 2020, 25, 100641.	1.1	8
29	Opa1 Overexpression Protects from Early-Onset Mpv17â^'/â^'-Related Mouse Kidney Disease. Molecular Therapy, 2020, 28, 1918-1930.	8.2	9
30	RCC1L (WBSCR16) isoforms coordinate mitochondrial ribosome assembly through their interaction with GTPases. PLoS Genetics, 2020, 16, e1008923.	3.5	18
31	Biallelic mutations in NDUFA8 cause complex I deficiency in two siblings with favorable clinical evolution. Molecular Genetics and Metabolism, 2020, 131, 349-357.	1.1	6
32	A Single Intravenous Injection of AAV-PHP.B-hNDUFS4 Ameliorates the Phenotype of Ndufs4 Mice. Molecular Therapy - Methods and Clinical Development, 2020, 17, 1071-1078.	4.1	32
33	Strategies for fighting mitochondrial diseases. Journal of Internal Medicine, 2020, 287, 665-684.	6.0	47
34	Awareness of rare and genetic neurological diseases among italian neurologist. A national survey. Neurological Sciences, 2020, 41, 1567-1570.	1.9	2
35	A homozygous MRPL24 mutation causes a complex movement disorder and affects the mitoribosome assembly. Neurobiology of Disease, 2020, 141, 104880.	4.4	29
36	ATPase Domain <scp>AFG3L2</scp> Mutations Alter <scp>OPA1</scp> Processing and Cause Optic Neuropathy. Annals of Neurology, 2020, 88, 18-32.	5.3	31

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37	Expanding the molecular and phenotypic spectrum of truncating MT-ATP6 mutations. Neurology: Genetics, 2020, 6, e381.	1.9	21
38	Impaired Mitochondrial ATP Production Downregulates Wnt Signaling via ER Stress Induction. Cell Reports, 2019, 28, 1949-1960.e6.	6.4	56
39	Knockdown of APOPT1/COA8 Causes Cytochrome c Oxidase Deficiency, Neuromuscular Impairment, and Reduced Resistance to Oxidative Stress in Drosophila melanogaster. Frontiers in Physiology, 2019, 10, 1143.	2.8	19
40	Breathe: Your Mitochondria Will Do the Restâ€¦ If They Are Healthy!. Cell Metabolism, 2019, 30, 628-629.	16.2	4
41	Experimental Therapies. , 2019, , 357-370.		0
42	Inhibition of proteasome rescues a pathogenic variant of respiratory chain assembly factor COA7. EMBO Molecular Medicine, 2019, 11, .	6.9	59
43	Lipomatosis Incidence and Characteristics in an Italian Cohort of Mitochondrial Patients. Frontiers in Neurology, 2019, 10, 160.	2.4	19
44	miRâ€181a/b downregulation exerts a protective action on mitochondrial disease models. EMBO Molecular Medicine, 2019, 11, .	6.9	58
45	APOPT 1/ COA 8 assists COX assembly and is oppositely regulated by UPS and ROS. EMBO Molecular Medicine, 2019, 11, .	6.9	19
46	RNase H1 directs origin-specific initiation of DNA replication in human mitochondria. PLoS Genetics, 2019, 15, e1007781.	3.5	58
47	RNase H1 Regulates Mitochondrial Transcription and Translation via the Degradation of 7S RNA. Frontiers in Genetics, 2019, 10, 1393.	2.3	12
48	Long-Term Sustained Effect of Liver-Targeted Adeno-Associated Virus Gene Therapy for Mitochondrial Neurogastrointestinal Encephalomyopathy. Human Gene Therapy, 2018, 29, 708-718.	2.7	39
49	Neuronal complex I deficiency occurs throughout the Parkinsonâ€™s disease brain, but is not associated with neurodegeneration or mitochondrial DNA damage. Acta Neuropathologica, 2018, 135, 409-425.	7.7	89
50	SURF1 knockout cloned pigs: Early onset of a severe lethal phenotype. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 2131-2142.	3.8	24
51	Cavitating Leukoencephalopathy With Posterior Predominance Caused by a Deletion in the APOPT1 Gene in an Indian Boy. Journal of Child Neurology, 2018, 33, 428-431.	1.4	16
52	Compound heterozygous missense and deep intronic variants in NDUFAF6 unraveled by exome sequencing and mRNA analysis. Journal of Human Genetics, 2018, 63, 563-568.	2.3	15
53	Mitochondrial complex III Rieske Fe-S protein processing and assembly. Cell Cycle, 2018, 17, 681-687.	2.6	70
54	MITOCHONDRIAL DISEASES I (Oral). Neuromuscular Disorders, 2018, 28, S87.	0.6	0

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55	P12â€...A novel model of cardiomyopathy reveals a tissue specific role for the complex i assembly factor ecsit. , 2018, , .		0
56	Genome editing in mitochondria corrects a pathogenic mtDNA mutation in vivo. Nature Medicine, 2018, 24, 1691-1695.	30.7	215
57	Rapamycin rescues mitochondrial myopathy via coordinated activation of autophagy and lysosomal biogenesis. EMBO Molecular Medicine, 2018, 10, .	6.9	86
58	Unravelling the Effects of the Mutation m.3571insC/MT-ND1 on Respiratory Complexes Structural Organization. International Journal of Molecular Sciences, 2018, 19, 764.	4.1	13
59	Mutations in TIMM50 compromise cell survival in OxPhosâ€dependent metabolic conditions. EMBO Molecular Medicine, 2018, 10, .	6.9	23
60	Mitochondrial PITRM1 peptidase loss-of-function in childhood cerebellar atrophy. Journal of Medical Genetics, 2018, 55, 599-606.	3.2	26
61	Human diseases associated with defects in assembly of OXPHOS complexes. Essays in Biochemistry, 2018, 62, 271-286.	4.7	75
62	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. Orphanet Journal of Rare Diseases, 2018, 13, 120.	2.7	61
63	A two-nuclease pathway involving RNase H1 is required for primer removal at human mitochondrial OriL. Nucleic Acids Research, 2018, 46, 9471-9483.	14.5	25
64	Perturbed Redox Signaling Exacerbates a Mitochondrial Myopathy. Cell Metabolism, 2018, 28, 764-775.e5.	16.2	70
65	Pure myopathy with enlarged mitochondria associated to a new mutation in MTND2 gene. Molecular Genetics and Metabolism Reports, 2017, 10, 24-27.	1.1	8
66	Novel mutation in mitochondrial Elongation Factor EF-Tu associated to dysplastic leukoencephalopathy and defective mitochondrial DNA translation. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2017, 1863, 961-967.	3.8	12
67	Down-regulation of the mitochondrial aspartate-glutamate carrier isoform 1 AGC1 inhibits proliferation and N-acetylaspartate synthesis in Neuro2A cells. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2017, 1863, 1422-1435.	3.8	22
68	MR-1S Interacts with PET100 and PET117 in Module-Based Assembly of Human Cytochrome c Oxidase. Cell Reports, 2017, 18, 1727-1738.	6.4	86
69	Paradoxical Inhibition of Glycolysis by Pioglitazone Opposes the Mitochondriopathy Caused by AIF Deficiency. EBioMedicine, 2017, 17, 75-87.	6.1	15
70	Clinicopathologic and molecular spectrum of RNASEH1 -related mitochondrial disease. Neurology: Genetics, 2017, 3, e149.	1.9	19
71	Recessive mutations in MSTO1 cause mitochondrial dynamics impairment, leading to myopathy and ataxia. Human Mutation, 2017, 38, 970-977.	2.5	44
72	MtDNAâ€maintenance defects: syndromes and genes. Journal of Inherited Metabolic Disease, 2017, 40, 587-599.	3.6	145

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73	Transcription Factor EB Controls Metabolic Flexibility during Exercise. Cell Metabolism, 2017, 25, 182-196.	16.2	250
74	Dysregulated mitophagy and mitochondrial organization in optic atrophy due to OPA1 mutations. Neurology, 2017, 88, 131-142.	1.1	81
75	A novel de novo dominant mutation inISCUassociated with mitochondrial myopathy. Journal of Medical Genetics, 2017, 54, 815-824.	3.2	25
76	International Workshop:. Neuromuscular Disorders, 2017, 27, 1126-1137.	0.6	58
77	PGD for the m.14487 T>C mitochondrial DNA mutation resulted in the birth of a healthy boy. Human Reproduction, 2017, 32, 698-703.	0.9	17
78	AAV9-based gene therapy partially ameliorates the clinical phenotype of a mouse model of Leigh syndrome. Gene Therapy, 2017, 24, 661-667.	4.5	50
79	Recessive mutations in novel gene MST01 cause early onset neuromuscular condition. Neuromuscular Disorders, 2017, 27, S176.	0.6	0
80	Revisiting mitochondrial ocular myopathies: a study from the Italian Network. Journal of Neurology, 2017, 264, 1777-1784.	3.6	32
81	TTC19 Plays a Husbandry Role on UQCRFS1 Turnover in the Biogenesis of Mitochondrial Respiratory Complex III. Molecular Cell, 2017, 67, 96-105.e4.	9.7	64
82	Defective mitochondrial rRNA methyltransferase MRM2 causes MELAS-like clinical syndrome. Human Molecular Genetics, 2017, 26, 4257-4266.	2.9	63
83	A Myopathy, Lactic Acidosis, Sideroblastic Anemia (Mlasa) Case Due to A Novel Pus1 Mutation. Turkish Journal of Haematology, 2017, 34, 376-377.	0.5	6
84	Quantitative proteomics suggests metabolic reprogramming during ETHE1 deficiency. Proteomics, 2016, 16, 1166-1176.	2.2	12
85	Myoclonus epilepsy in mitochondrial disorders. Epileptic Disorders, 2016, 18, 94-102.	1.3	22
86	COA7(C1orf163/RESA1) mutations associated with mitochondrial leukoencephalopathy and cytochrome c oxidase deficiency. Journal of Medical Genetics, 2016, 53, 846-849.	3.2	40
87	Tissue- and species-specific differences in cytochrome c oxidase assembly induced by SURF1 defects. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2016, 1862, 705-715.	3.8	21
88	Data on cytochrome c oxidase assembly in mice and human fibroblasts or tissues induced by SURF1 defect. Data in Brief, 2016, 7, 1004-1009.	1.0	1
89	Exome sequencing coupled with mRNA analysis identifies NDUFAF6 as a Leigh gene. Molecular Genetics and Metabolism, 2016, 119, 214-222.	1.1	21
90	Recurrent De Novo and Biallelic Variation of ATAD3A , Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. American Journal of Human Genetics, 2016, 99, 831-845.	6.2	146

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91	Mitochondrial medicine: Disease genes and mechanisms. Biochimica Et Biophysica Acta - Bioenergetics, 2016, 1857, e6.	1.0	0
92	Mitochondrial Matchmaking. New England Journal of Medicine, 2016, 375, 1894-1896.	27.0	8
93	Mitochondrial diseases. Nature Reviews Disease Primers, 2016, 2, 16080.	30.5	1,001
94	FGF21 is a biomarker for mitochondrial translation and mtDNA maintenance disorders. Neurology, 2016, 87, 2290-2299.	1.1	167
95	Defective <scp>PITRM</scp> 1 mitochondrial peptidase is associated with Aî² amyloidotic neurodegeneration. EMBO Molecular Medicine, 2016, 8, 176-190.	6.9	60
96	Mitochondrial Genes and Neurodegenerative Disease. , 2016, , 81-106.		1
97	Disease-Causing SDHAF1 Mutations Impair Transfer of Fe-S Clusters to SDHB. Cell Metabolism, 2016, 23, 292-302.	16.2	89
98	â€œMitochondrial neuropathiesâ€: A survey from the large cohort of the Italian Network. Neuromuscular Disorders, 2016, 26, 272-276.	0.6	37
99	Liver transplant in ethylmalonic encephalopathy: a new treatment for an otherwise fatal disease. Brain, 2016, 139, 1045-1051.	7.6	65
100	New genes and pathomechanisms in mitochondrial disorders unraveled by NGS technologies. Biochimica Et Biophysica Acta - Bioenergetics, 2016, 1857, 1326-1335.	1.0	87
101	Mitochondrial DNA sequence characteristics modulate the size of the genetic bottleneck. Human Molecular Genetics, 2016, 25, 1031-1041.	2.9	53
102	Clinical and genetic features of paroxysmal kinesigenic dyskinesia in Italian patients. European Journal of Paediatric Neurology, 2016, 20, 152-157.	1.6	14
103	Reduced mitochondrial Ca2+ transients stimulate autophagy in human fibroblasts carrying the 13514A>G mutation of the ND5 subunit of NADH dehydrogenase. Cell Death and Differentiation, 2016, 23, 231-241.	11.2	51
104	A nonsense mutation of human <scp>XRCC</scp> 4 is associated with adultâ€onset progressive encephalocardiomyopathy. EMBO Molecular Medicine, 2015, 7, 918-929.	6.9	24
105	The Opa1-Dependent Mitochondrial Cristae Remodeling Pathway Controls Atrophic, Apoptotic, and Ischemic Tissue Damage. Cell Metabolism, 2015, 21, 834-844.	16.2	350
106	COQ4 Mutations Cause a Broad Spectrum of Mitochondrial Disorders Associated with CoQ10 Deficiency. American Journal of Human Genetics, 2015, 96, 309-317.	6.2	86
107	Emerging concepts in the therapy of mitochondrial disease. Biochimica Et Biophysica Acta - Bioenergetics, 2015, 1847, 544-557.	1.0	96
108	RNASEH1 Mutations Impair mtDNA Replication and Cause Adult-Onset Mitochondrial Encephalomyopathy. American Journal of Human Genetics, 2015, 97, 186-193.	6.2	91

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109	Opa1 Overexpression Ameliorates the Phenotype of Two Mitochondrial Disease Mouse Models. Cell Metabolism, 2015, 21, 845-854.	16.2	202
110	Distributed abnormalities of brain white matter architecture in patients with dominant optic atrophy and OPA1 mutations. Journal of Neurology, 2015, 262, 1216-1227.	3.6	5
111	Syndromic parkinsonism and dementia associated with <scp>OPA</scp>1 missense mutations. Annals of Neurology, 2015, 78, 21-38.	5.3	154
112	Nuclear gene mutations as the cause of mitochondrial complex III deficiency. Frontiers in Genetics, 2015, 6, 134.	2.3	116
113	Redefining phenotypes associated with mitochondrial DNA single deletion. Journal of Neurology, 2015, 262, 1301-1309.	3.6	68
114	Expanding the Clinical and Magnetic Resonance Spectrum of Leukoencephalopathy with Thalamus and Brainstem Involvement and High Lactate (LTBL) in a Patient Harboring a Novel EARS2 Mutation. JIMD Reports, 2015, 23, 85-89.	1.5	15
115	Mutations in NDUFB11, Encoding a Complex I Component of the Mitochondrial Respiratory Chain, Cause Microphthalmia with Linear Skin Defects Syndrome. American Journal of Human Genetics, 2015, 96, 640-650.	6.2	56
116	Severe early onset ethylmalonic encephalopathy with West syndrome. Metabolic Brain Disease, 2015, 30, 1537-1545.	2.9	13
117	Loss of apoptosis-inducing factor critically affects MIA40 function. Cell Death and Disease, 2015, 6, e1814-e1814.	6.3	77
118	Foxg1 localizes to mitochondria and coordinates cell differentiation and bioenergetics. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 13910-13915.	7.1	54
119	Dysregulated mitophagy and mitochondrial transport in sensori-motor neuropathy due to â€œDominant Optic Atrophyâ€•plus with OPA1 (Optic Atrophy 1) mutations. Neuromuscular Disorders, 2015, 25, S185-S186.	0.6	0
120	Mitochondrial myopathy biomarker Fibroblast growth factor 21 is induced by muscle mtDNA instability and translation defects. Mitochondrion, 2015, 24, S45-S46.	3.4	3
121	A family with paroxysmal nonkinesigenic dyskinesias (PNKD): Evidence of mitochondrial dysfunction. European Journal of Paediatric Neurology, 2015, 19, 64-68.	1.6	13
122	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
123	A Novel Homozygous YARS2 Mutation in Two Italian Siblings and a Review of Literature. JIMD Reports, 2014, 20, 95-101.	1.5	19
124	Functional Characterization of drim2, the Drosophila melanogaster Homolog of the Yeast Mitochondrial Deoxynucleotide Transporter. Journal of Biological Chemistry, 2014, 289, 7448-7459.	3.4	13
125	Leigh Syndrome in Drosophila melanogaster. Journal of Biological Chemistry, 2014, 289, 29235-29246.	3.4	22
126	Common and Novel TMEM70 Mutations in a Cohort of Italian Patients with Mitochondrial Encephalocardiomyopathy. JIMD Reports, 2014, 15, 71-8.	1.5	23

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127	Cavitating leukoencephalopathy with multiple mitochondrial dysfunction syndrome and NFU1 mutations. Frontiers in Genetics, 2014, 5, 412.	2.3	49
128	The impairment of HCCS leads to MLS syndrome by activating a nonâ€canonical cell death pathway in the brain and eyes. EMBO Molecular Medicine, 2014, 6, 849-849.	6.9	0
129	Mutations in APOPT1, Encoding a Mitochondrial Protein, Cause Cavitating Leukoencephalopathy with Cytochrome c Oxidase Deficiency. American Journal of Human Genetics, 2014, 95, 315-325.	6.2	64
130	NAD+-Dependent Activation of Sirt1 Corrects the Phenotype in a Mouse Model of Mitochondrial Disease. Cell Metabolism, 2014, 19, 1042-1049.	16.2	293
131	Gene Therapy Using a Liver-targeted AAV Vector Restores Nucleoside and Nucleotide Homeostasis in a Murine Model of MNGIE. Molecular Therapy, 2014, 22, 901-907.	8.2	55
132	VARS2andTARS2Mutations in Patients with Mitochondrial Encephalomyopathies. Human Mutation, 2014, 35, 983-989.	2.5	86
133	Pharmacological Inhibition of Poly(ADP-Ribose) Polymerases Improves Fitness and Mitochondrial Function in Skeletal Muscle. Cell Metabolism, 2014, 19, 1034-1041.	16.2	211
134	The m.3243A>G mitochondrial DNA mutation and related phenotypes. A matter of gender?. Journal of Neurology, 2014, 261, 504-510.	3.6	119
135	AAV-mediated Liver-specific MPV17 Expression Restores mtDNA Levels and Prevents Diet-induced Liver Failure. Molecular Therapy, 2014, 22, 10-17.	8.2	47
136	Efficient mitochondrial biogenesis drives incomplete penetrance in Leberâ€™s hereditary optic neuropathy. Brain, 2014, 137, 335-353.	7.6	229
137	The isolated carboxyâ€terminal domain of human mitochondrial leucylâ€<scp>tRNA</scp> synthetase rescues the pathological phenotype of mitochondrial <scp>tRNA</scp> mutations in human cells. EMBO Molecular Medicine, 2014, 6, 169-182.	6.9	43
138	A SIRT7-Dependent Acetylation Switch of GABPÎ²1 Controls Mitochondrial Function. Cell Metabolism, 2014, 20, 856-869.	16.2	214
139	Complex IV-deficient Surf1â^'/â^' mice initiate mitochondrial stress responses. Biochemical Journal, 2014, 462, 359-371.	3.7	89
140	Myoclonus in mitochondrial disorders. Movement Disorders, 2014, 29, 722-728.	3.9	33
141	Novel (ovario) leukodystrophy related to AARS2 mutations. Neurology, 2014, 82, 2063-2071.	1.1	172
142	Early Macular Retinal Ganglion Cell Loss in Dominant Optic Atrophy: Genotype-Phenotype Correlation. American Journal of Ophthalmology, 2014, 158, 628-636.e3.	3.3	56
143	UCP4C mediates uncoupled respiration in larvae of Drosophila melanogaster. EMBO Reports, 2014, 15, 586-591.	4.5	31
144	Mitochondrial Diseases in Childhood. Current Molecular Medicine, 2014, 14, 1069-1078.	1.3	3

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145	Ethylmalonic Encephalopathy. , 2014, , 157-163.		0
146	Adult-onset Alexander disease, associated with a mutation in an alternative GFAP transcript, may be phenotypically modulated by a non-neutral HDAC6 variant. Orphanet Journal of Rare Diseases, 2013, 8, 66.	2.7	21
147	Adult-onset leukodystrophies from respiratory chain disorders: do they exist?. Journal of Neurology, 2013, 260, 1617-1623.	3.6	5
148	Improved insulin sensitivity associated with reduced mitochondrial complex IV assembly and activity. FASEB Journal, 2013, 27, 1371-1380.	0.5	29
149	Mutations in FBXL4, Encoding a Mitochondrial Protein, Cause Early-Onset Mitochondrial Encephalomyopathy. American Journal of Human Genetics, 2013, 93, 482-495.	6.2	138
150	Peripheral neuropathy in mitochondrial disorders. Lancet Neurology, The, 2013, 12, 1011-1024.	10.2	101
151	MTO1 Mutations are Associated with Hypertrophic Cardiomyopathy and Lactic Acidosis and Cause Respiratory Chain Deficiency in Humans and Yeast. Human Mutation, 2013, 34, 1501-1509.	2.5	67
152	Decreased in vitro Mitochondrial Function is Associated with Enhanced Brain Metabolism, Blood Flow, and Memory in Surfl-Deficient Mice. Journal of Cerebral Blood Flow and Metabolism, 2013, 33, 1605-1611.	4.3	35
153	Altered Sulfide (H2S) Metabolism in Ethylmalonic Encephalopathy. Cold Spring Harbor Perspectives in Biology, 2013, 5, a011437-a011437.	5.5	39
154	LYRM7/MZM1L is a UQCRFS1 chaperone involved in the last steps of mitochondrial Complex III assembly in human cells. Biochimica Et Biophysica Acta - Bioenergetics, 2013, 1827, 285-293.	1.0	71
155	Phenotypic heterogeneity of the 8344A>G mtDNA â€œMERRFâ€•mutation. Neurology, 2013, 80, 2049-2054.	1.1	157
156	New treatments for mitochondrial diseaseâ€”no time to drop our standards. Nature Reviews Neurology, 2013, 9, 474-481.	10.1	157
157	A case of Leber hereditary optic neuropathy plus dystonia caused by G14459A mitochondrial mutation. Neurological Sciences, 2013, 34, 407-408.	1.9	15
158	Phenylbutyrate Therapy for Pyruvate Dehydrogenase Complex Deficiency and Lactic Acidosis. Science Translational Medicine, 2013, 5, 175ra31.	12.4	59
159	SURF1 deficiency causes demyelinating Charcot-Marie-Tooth disease. Neurology, 2013, 81, 1523-1530.	1.1	53
160	Proteome adaptations in Ethe1-deficient mice indicate a role in lipid catabolism and cytoskeleton organization via post-translational protein modifications. Bioscience Reports, 2013, 33, .	2.4	31
161	A Homozygous Mutation in<scp>LYRM</scp>7/<scp>MZM</scp>1<scp>L</scp>Associated with Early Onset Encephalopathy, Lactic Acidosis, and Severe Reduction of Mitochondrial Complex<scp>III</scp>Activity. Human Mutation, 2013, 34, 1619-1622.	2.5	60
162	The impairment of HCCS leads to MLS syndrome by activating a nonâ€canonical cell death pathway in the brain and eyes. EMBO Molecular Medicine, 2013, 5, 280-293.	6.9	33

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163	Idebenone treatment in patients with OPA1-mutant dominant optic atrophy. Brain, 2013, 136, e231-e231.	7.6	62
164	Mutation screening of 75 candidate genes in 152 complex I deficiency cases identifies pathogenic variants in 16 genes includingNDUFB9. Journal of Medical Genetics, 2012, 49, 83-89.	3.2	78
165	Leukoencephalopathy with thalamus and brainstem involvement and high lactate â€˜LTBLâ€™ caused by EARS2 mutations. Brain, 2012, 135, 1387-1394.	7.6	187
166	Hepatocerebral form of mitochondrial DNA depletion syndrome due to mutation in MPV17 gene. Saudi Journal of Gastroenterology, 2012, 18, 285.	1.1	18
167	A novel homozygous mutation in SUCLA2 gene identified by exome sequencing. Molecular Genetics and Metabolism, 2012, 107, 403-408.	1.1	38
168	Molecular diagnosis in mitochondrial complex I deficiency using exome sequencing. Journal of Medical Genetics, 2012, 49, 277-283.	3.2	182
169	Cowchock Syndrome Is Associated with a Mutation in Apoptosis-Inducing Factor. American Journal of Human Genetics, 2012, 91, 1095-1102.	6.2	134
170	Partial tandem duplication of mtDNAâ€“tRNAPhe impairs mtDNA translation in late-onset mitochondrial myopathy. Neuromuscular Disorders, 2012, 22, 50-55.	0.6	2
171	Microscale oxygraphy reveals OXPHOS impairment in MRC mutant cells. Mitochondrion, 2012, 12, 328-335.	3.4	90
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