Massimo Zeviani

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/6734509/publications.pdf

Version: 2024-02-01

491 papers	46,207 citations	1531 109 h-index	3100 193 g-index
518 all docs	518 docs citations	518 times ranked	32170 citing authors

#	Article	IF	Citations
1	Identification and characterization of a spinal muscular atrophy-determining gene. Cell, 1995, 80, 155-165.	13.5	3,424
2	Mitochondrial DNA Deletions in Progressive External Ophthalmoplegia and Kearns-Sayre Syndrome. New England Journal of Medicine, 1989, 320, 1293-1299.	13.9	1,012
3	Mitochondrial diseases. Nature Reviews Disease Primers, 2016, 2, 16080.	18.1	1,001
4	Human mitochondrial DNA deletions associated with mutations in the gene encoding Twinkle, a phage T7 gene 4-like protein localized in mitochondria. Nature Genetics, 2001, 28, 223-231.	9.4	803
5	Spastic Paraplegia and OXPHOS Impairment Caused by Mutations in Paraplegin, a Nuclear-Encoded Mitochondrial Metalloprotease. Cell, 1998, 93, 973-983.	13.5	784
6	Mitochondrial myopathies. Annals of Neurology, 1985, 17, 521-538.	2.8	762
7	Oxygen sensing requires mitochondrial ROS but not oxidative phosphorylation. Cell Metabolism, 2005, 1, 409-414.	7.2	678
8	An autosomal dominant disorder with multiple deletions of mitochondrial DNA starting at the D-loop region. Nature, 1989, 339, 309-311.	13.7	640
9	Deletions of mitochondrial DNA in Kearnsâ€Sayre syndrome. Neurology, 1988, 38, 1339-1339.	1.5	624
10	Role of Adenine Nucleotide Translocator 1 in mtDNA Maintenance. Science, 2000, 289, 782-785.	6.0	591
11	A direct repeat is a hotspot for large-scale deletion of human mitochondrial DNA. Science, 1989, 244, 346-349.	6.0	566
12	Familial Progressive Sensorineural Deafness Is Mainly Due to the mtDNA A1555G Mutation and Is Enhanced by Treatment with Aminoglycosides. American Journal of Human Genetics, 1998, 62, 27-35.	2.6	504
13	Mutations of SURF-1 in Leigh Disease Associated with Cytochrome c Oxidase Deficiency. American Journal of Human Genetics, 1998, 63, 1609-1621.	2.6	504
14	Deletions of mitochondrial DNA in Kearnsâ€Sayre syndrome. Neurology, 1988, 38, 1339-1339.	1.5	445
15	The Molecular Dissection of mtDNA Haplogroup H Confirms That the Franco-Cantabrian Glacial Refuge Was a Major Source for the European Gene Pool. American Journal of Human Genetics, 2004, 75, 910-918.	2.6	397
16	Phenotypic spectrum associated with mutations of the mitochondrial polymerase Â gene. Brain, 2006, 129, 1674-1684.	3.7	397
17	Maternally inherited myopathy and cardiomyopathy: association with mutation in mitochondrial DNA tRNALeu(UUR). Lancet, The, 1991, 338, 143-147.	6.3	395
18	Cytochromec oxidase deficiency in leigh syndrome. Annals of Neurology, 1987, 22, 498-506.	2.8	390

#	Article	IF	Citations
19	Multi-system neurological disease is common in patients with OPA1 mutations. Brain, 2010, 133, 771-786.	3.7	385
20	MPV17 encodes an inner mitochondrial membrane protein and is mutated in infantile hepatic mitochondrial DNA depletion. Nature Genetics, 2006, 38, 570-575.	9.4	380
21	Mitochondrial disorders. Brain, 2004, 127, 2153-2172.	3.7	362
22	Loss of ETHE1, a mitochondrial dioxygenase, causes fatal sulfide toxicity in ethylmalonic encephalopathy. Nature Medicine, 2009, 15, 200-205.	15.2	358
23	The Opa1-Dependent Mitochondrial Cristae Remodeling Pathway Controls Atrophic, Apoptotic, and Ischemic Tissue Damage. Cell Metabolism, 2015, 21, 834-844.	7.2	350
24	The spectrum of clinical disease caused by the A467T and W748S POLG mutations: a study of 26 cases. Brain, 2006, 129, 1685-1692.	3.7	337
25	Clinical Expression of Leber Hereditary Optic Neuropathy Is Affected by the Mitochondrial DNAâ€“Haplogroup Background. American Journal of Human Genetics, 2007, 81, 228-233.	2.6	331
26	De novo and inherited deletions of the 5q13 region in spinal muscular atrophies. Science, 1994, 264, 1474-1477.	6.0	330
27	Haplotype and phylogenetic analyses suggest that one European-specific mtDNA background plays a role in the expression of Leber hereditary optic neuropathy by increasing the penetrance of the primary mutations 11778 and 14484. American Journal of Human Genetics, 1997, 60, 1107-21.	2.6	326
28	Systematic identification of human mitochondrial disease genes through integrative genomics. Nature Genetics, 2006, 38, 576-582.	9.4	321
29	Transthyretin. Neurology, 1986, 36, 900-900.	1.5	300
30	NAD+-Dependent Activation of Sirt1 Corrects the Phenotype in a Mouse Model of Mitochondrial Disease. Cell Metabolism, 2014, 19, 1042-1049.	7.2	293
31	Infantile hepatocerebral syndromes associated with mutations in the mitochondrial DNA polymerase-ÂA. Brain, 2005, 128, 723-731.	3.7	284
32	Increased longevity and refractoriness to Ca2+-dependent neurodegeneration in Surf1 knockout mice. Human Molecular Genetics, 2007, 16, 431-444.	1.4	279
33	An autosomal locus predisposing to deletions of mitochondrial DNA. Nature Genetics, 1995, 9, 146-151.	9.4	268
34	Mutations of mitochondrial DNA polymerase ?A are a frequent cause of autosomal dominant or recessive progressive external ophthalmoplegia. Annals of Neurology, 2002, 52, 211-219.	2.8	257
35	Cytochromec Oxidase subunit I microdeletion in a patient with motor neuron disease. Annals of Neurology, 1998, 43, 110-116.	2.8	251
36	Transcription Factor EB Controls Metabolic Flexibility during Exercise. Cell Metabolism, 2017, 25, 182-196.	7.2	250

#	Article	IF	Citations
37	Autosomal recessive mitochondrial ataxic syndrome due to mitochondrial polymerase Î³ mutations. Neurology, 2005, 64, 1204-1208.	1.5	246
38	In Vivo Correction of COX Deficiency by Activation of the AMPK/PGC-1Î± Axis. Cell Metabolism, 2011, 14, 80-90.	7.2	245
39	Exome sequencing identifies ACAD9 mutations as a cause of complex I deficiency. Nature Genetics, 2010, 42, 1131-1134.	9.4	234
40	SDHAF1, encoding a LYR complex-II specific assembly factor, is mutated in SDH-defective infantile leukoencephalopathy. Nature Genetics, 2009, 41, 654-656.	9.4	233
41	Clinical and molecular findings in children with complex I deficiency. Biochimica Et Biophysica Acta - Bioenergetics, 2004, 1659, 136-147.	0.5	231
42	Efficient mitochondrial biogenesis drives incomplete penetrance in Leberâ€™s hereditary optic neuropathy. Brain, 2014, 137, 335-353.	3.7	229
43	Maternally inherited hearing loss, ataxia and myoclonus associated with a novel point mutation in mitochondrial tRNA Ser(UCN) gene. Human Molecular Genetics, 1995, 4, 1421-1427.	1.4	218
44	Genome editing in mitochondria corrects a pathogenic mtDNA mutation in vivo. Nature Medicine, 2018, 24, 1691-1695.	15.2	215
45	A SIRT7-Dependent Acetylation Switch of GABPÎ²1 Controls Mitochondrial Function. Cell Metabolism, 2014, 20, 856-869.	7.2	214
46	Pharmacological Inhibition of Poly(ADP-Ribose) Polymerases Improves Fitness and Mitochondrial Function in Skeletal Muscle. Cell Metabolism, 2014, 19, 1034-1041.	7.2	211
47	Mitochondrial medicine: A metabolic perspective on the pathology of oxidative phosphorylation disorders. Cell Metabolism, 2006, 3, 9-13.	7.2	210
48	Opa1 Overexpression Ameliorates the Phenotype of Two Mitochondrial Disease Mouse Models. Cell Metabolism, 2015, 21, 845-854.	7.2	202
49	Risk of developing a mitochondrial DNA deletion disorder. Lancet, The, 2004, 364, 592-596.	6.3	201
50	Severe X-Linked Mitochondrial Encephalomyopathy Associated with a Mutation in Apoptosis-Inducing Factor. American Journal of Human Genetics, 2010, 86, 639-649.	2.6	199
51	Mitochondrial DNA haplogroup K is associated with a lower risk of Parkinson's disease in Italians. European Journal of Human Genetics, 2005, 13, 748-752.	1.4	197
52	Ethylmalonic Encephalopathy Is Caused by Mutations in ETHE1, a Gene Encoding a Mitochondrial Matrix Protein. American Journal of Human Genetics, 2004, 74, 239-252.	2.6	192
53	Idebenone Treatment In Leber's Hereditary Optic Neuropathy. Brain, 2011, 134, e188-e188.	3.7	192
54	Lack of the Mitochondrial Protein Acylglycerol Kinase Causes Sengers Syndrome. American Journal of Human Genetics, 2012, 90, 314-320.	2.6	192

#	Article	IF	Citations
55	Evidence that nebulin is a protein-ruler in muscle thin filaments. FEBS Letters, 1991, 282, 313-316.	1.3	187
56	Leukoencephalopathy with thalamus and brainstem involvement and high lactate â€˜LTBLâ€™ caused by EARS2 mutations. Brain, 2012, 135, 1387-1394.	3.7	187
57	MELAS syndrome. Neurology, 1988, 38, 751-751.	1.5	185
58	Assembly of the oxidative phosphorylation system in humans: What we have learned by studying its defects. Biochimica Et Biophysica Acta - Molecular Cell Research, 2009, 1793, 200-211.	1.9	182
59	Molecular diagnosis in mitochondrial complex I deficiency using exome sequencing. Journal of Medical Genetics, 2012, 49, 277-283.	1.5	182
60	Identification of an X-Chromosomal Locus and Haplotype Modulating the Phenotype of a Mitochondrial DNA Disorder. American Journal of Human Genetics, 2005, 77, 1086-1091.	2.6	181
61	Identification of the gene encoding the human mitochondrial RNA polymerase (h-mtRPOL) by cyberscreening of the Expressed Sequence Tags database. Human Molecular Genetics, 1997, 6, 615-625.	1.4	178
62	POLG1 mutations cause a syndromic epilepsy with occipital lobe predilection. Brain, 2008, 131, 818-828.	3.7	176
63	Mutational spectrum of the CHAC gene in patients with chorea-acanthocytosis. European Journal of Human Genetics, 2002, 10, 773-781.	1.4	172
64	Infantile Encephalopathy and Defective Mitochondrial DNA Translation in Patients with Mutations of Mitochondrial Elongation Factors EFG1 and EFTu. American Journal of Human Genetics, 2007, 80, 44-58.	2.6	172
65	Novel (ovario) leukodystrophy related to AARS2 mutations. Neurology, 2014, 82, 2063-2071.	1.5	172
66	FGF21 is a biomarker for mitochondrial translation and mtDNA maintenance disorders. Neurology, 2016, 87, 2290-2299.	1.5	167
67	Haplogroup Effects and Recombination of Mitochondrial DNA: Novel Clues from the Analysis of Leber Hereditary Optic Neuropathy Pedigrees. American Journal of Human Genetics, 2006, 78, 564-574.	2.6	166
68	Complete loss-of-function of the heart/muscle-specific adenine nucleotide translocator is associated with mitochondrial myopathy and cardiomyopathy. Human Molecular Genetics, 2005, 14, 3079-3088.	1.4	165
69	Severe Infantile Encephalomyopathy Caused by a Mutation in COX6B1, a Nucleus-Encoded Subunit of Cytochrome C Oxidase. American Journal of Human Genetics, 2008, 82, 1281-1289.	2.6	165
70	Mutations of the Mitochondrial-tRNA Modifier MTO1 Cause Hypertrophic Cardiomyopathy and Lactic Acidosis. American Journal of Human Genetics, 2012, 90, 1079-1087.	2.6	164
71	Clinical and molecular features of mitochondrial DNA depletion syndromes. Journal of Inherited Metabolic Disease, 2009, 32, 143-158.	1.7	161
72	Impaired complex III assembly associated with BCS1L gene mutations in isolated mitochondrial encephalopathy. Human Molecular Genetics, 2007, 16, 1241-1252.	1.4	158

#	Article	IF	Citations
73	Isolation of mitochondria for biogenetical studies: An update. Mitochondrion, 2010, 10, 253-262.	1.6	158
74	Phenotypic heterogeneity of the 8344A>G mtDNA â€œMERRFâ€•mutation. Neurology, 2013, 80, 2049-2054.	1.5	157
75	New treatments for mitochondrial diseaseâ€”no time to drop our standards. Nature Reviews Neurology, 2013, 9, 474-481.	4.9	157
76	Expression of the Ciona intestinalis Alternative Oxidase (AOX) in Drosophila Complements Defects in Mitochondrial Oxidative Phosphorylation. Cell Metabolism, 2009, 9, 449-460.	7.2	156
77	Syndromic parkinsonism and dementia associated with <scp>OPA</scp>1 missense mutations. Annals of Neurology, 2015, 78, 21-38.	2.8	154
78	Fatal infantile cytochrome c oxidase deficiency. Neurology, 1985, 35, 802-802.	1.5	149
79	Mutations in TTC19 cause mitochondrial complex III deficiency and neurological impairment in humans and flies. Nature Genetics, 2011, 43, 259-263.	9.4	148
80	Mutations of ANT1 , Twinkle , and POLG1 in sporadic progressive external ophthalmoplegia (PEO). Neurology, 2003, 60, 1354-1356.	1.5	147
81	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.	2.6	146
82	An international classification of inherited metabolic disorders (<scp>ICIMD</scp>). Journal of Inherited Metabolic Disease, 2021, 44, 164-177.	1.7	146
83	MtDNAâ€maintenance defects: syndromes and genes. Journal of Inherited Metabolic Disease, 2017, 40, 587-599.	1.7	145
84	Identification and Characterization of Human cDNAs Specific to BCS1, PET112, SCO1, COX15, and COX11, Five Genes Involved in the Formation and Function of the Mitochondrial Respiratory Chain. Genomics, 1998, 54, 494-504.	1.3	144
85	Epileptic phenotypes associated with mitochondrial disorders. Neurology, 2001, 56, 1340-1346.	1.5	143
86	A MERRF/MELAS Overlap Syndrome Associated with a New Point Mutation in the Mitochondrial DNA tRNA^Lys Gene. European Journal of Human Genetics, 1993, 1, 80-87.	1.4	143
87	Mutations in FBXL4, Encoding a Mitochondrial Protein, Cause Early-Onset Mitochondrial Encephalomyopathy. American Journal of Human Genetics, 2013, 93, 482-495.	2.6	138
88	Combined treatment with oral metronidazole and N-acetylcysteine is effective in ethylmalonic encephalopathy. Nature Medicine, 2010, 16, 869-871.	15.2	136
89	Hereditary spastic paraplegia is a novel phenotype for GJA12/GJC2 mutations. Brain, 2009, 132, 426-438.	3.7	135
90	Characterization of SURF-1 Expression and Surf-1p Function in Normal and Disease Conditions. Human Molecular Genetics, 1999, 8, 2533-2540.	1.4	134

#	Article	IF	Citations
91	Cowchock Syndrome Is Associated with a Mutation in Apoptosis-Inducing Factor. American Journal of Human Genetics, 2012, 91, 1095-1102.	2.6	134
92	Mitochondrial disease associated with the T8993G mutation of the mitochondrial ATPase 6Âgene: a clinical, biochemical, and molecular study in six families. Journal of Neurology, Neurosurgery and Psychiatry, 1997, 63, 16-22.	0.9	133
93	Novel mutations of ND genes in complex I deficiency associated with mitochondrial encephalopathy. Brain, 2007, 130, 1894-1904.	3.7	131
94	Nonsense mutation in pseudouridylate synthase 1 (PUS1) in two brothers affected by myopathy, lactic acidosis and sideroblastic anaemia (MLASA). Journal of Medical Genetics, 2006, 44, 173-180.	1.5	128
95	Dysfunctions of Cellular Oxidative Metabolism in Patients with Mutations in the NDUFS1 and NDUFS4 Genes of Complex I. Journal of Biological Chemistry, 2006, 281, 10374-10380.	1.6	128
96	Human mitochondrial complex I assembly is mediated by NDUFAF1. FEBS Journal, 2005, 272, 5317-5326.	2.2	126
97	Expression of the SMN Gene, the Spinal Muscular Atrophy Determining Gene, in the Mammalian Central Nervous System. Human Molecular Genetics, 1997, 6, 1961-1971.	1.4	125
98	Structure-function defects of human mitochondrial DNA polymerase in autosomal dominant progressive external ophthalmoplegia. Nature Structural and Molecular Biology, 2004, 11, 770-776.	3.6	123
99	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.	2.6	123
100	Loss-of-function mutations of SURF-1 are specifically associated with Leigh syndrome with cytochromec oxidase deficiency. Annals of Neurology, 1999, 46, 161-166.	2.8	121
101	A nonsense mutation in the NDUFS4 gene encoding the 18 kDa (AQDQ) subunit of complex I abolishes assembly and activity of the complex in a patient with Leigh-like syndrome. Human Molecular Genetics, 2001, 10, 529-535.	1.4	120
102	Dominantly inherited mitochondrial myopathy with multiple deletions of mitochondrial DNA. Neurology, 1991, 41, 1053-1053.	1.5	120
103	The m.3243A>G mitochondrial DNA mutation and related phenotypes. A matter of gender?. Journal of Neurology, 2014, 261, 504-510.	1.8	119
104	Complex I deficiency is Associated with 3243G:C Mitochondrial DNA in Osteosarcoma Cell Cybrids. Human Molecular Genetics, 1996, 5, 123-129.	1.4	117
105	Mitochondrial disorders of the OXPHOS system. FEBS Letters, 2021, 595, 1062-1106.	1.3	117
106	A novel mtDNA mutation in the ND5 subunit of complex I in two MELAS patients. Annals of Neurology, 2001, 49, 106-110.	2.8	116
107	Assembly Factors of Human Mitochondrial Respiratory Chain Complexes: Physiology and Pathophysiology. Advances in Experimental Medicine and Biology, 2012, 748, 65-106.	0.8	116
108	Nuclear gene mutations as the cause of mitochondrial complex III deficiency. Frontiers in Genetics, 2015, 6, 134.	1.1	116

#	Article	IF	Citations
109	Genotype to phenotype correlations in mitochondrial encephalomyopathies associated with the A3243G mutation of mitochondrial DNA. Journal of Neurology, 1995, 242, 304-312.	1.8	115
110	Nucleus-driven multiple large-scale deletions of the human mitochondrial genome: a new autosomal dominant disease. American Journal of Human Genetics, 1990, 47, 904-14.	2.6	115
111	Myoclonic epilepsy and ragged-red fibers with cytochrome oxidase deficiency: Neuropathology, biochemistry, and molecular genetics. Annals of Neurology, 1989, 26, 20-33.	2.8	111
112	Mitochondrial DNA mutations in patients with postlingual, nonsyndromic hearing impairment. European Journal of Human Genetics, 2005, 13, 26-33.	1.4	110
113	Isolation of a cDNA clone encoding subunit IV of human cytochrome c oxidase. Gene, 1987, 55, 205-217.	1.0	109
114	Cloning of human and rat cDNAs encoding the mitochondrial single-stranded DNA-binding protein (SSB). Gene, 1993, 126, 219-225.	1.0	109
115	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. Human Molecular Genetics, 2000, 9, 2733-2742.	1.4	109
116	Disorders of nuclear-mitochondrial intergenomic signaling. Gene, 2005, 354, 162-168.	1.0	108
117	Rapid detection of the A----G(8344) mutation of mtDNA in Italian families with myoclonus epilepsy and ragged-red fibers (MERRF). American Journal of Human Genetics, 1991, 48, 203-11.	2.6	108
118	Mitochondrial disorders. Current Opinion in Neurology, 2007, 20, 564-571.	1.8	107
119	FASTKD2 Nonsense Mutation in an Infantile Mitochondrial Encephalomyopathy Associated with Cytochrome C Oxidase Deficiency. American Journal of Human Genetics, 2008, 83, 415-423.	2.6	107
120	Mutations in COX7B Cause Microphthalmia with Linear Skin Lesions, an Unconventional Mitochondrial Disease. American Journal of Human Genetics, 2012, 91, 942-949.	2.6	104
121	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.	3.5	102
122	Peripheral neuropathy in mitochondrial disorders. Lancet Neurology, The, 2013, 12, 1011-1024.	4.9	101
123	A novel nonsense mutation (Q352X) in the mitochondrial cytochrome b gene associated with a combined deficiency of complexes I and III. Neuromuscular Disorders, 2002, 12, 49-52.	0.3	100
124	Tissue distribution and transmission of mitochondrial DNA deletions in mitochondrial myopathies. Annals of Neurology, 1990, 28, 94-97.	2.8	98
125	Emerging concepts in the therapy of mitochondrial disease. Biochimica Et Biophysica Acta - Bioenergetics, 2015, 1847, 544-557.	0.5	96
126	GJA12 mutations in children with recessive hypomyelinating leukoencephalopathy. Neurology, 2006, 67, 273-279.	1.5	95

#	Article	IF	Citations
127	Neural stem cells traffic functional mitochondria via extracellular vesicles. PLoS Biology, 2021, 19, e3001166.	2.6	95
128	Single-stranded-DNA-binding proteins from human mitochondria and Escherichia coli have analogous physicochemical properties. FEBS Journal, 1994, 221, 435-443.	0.2	94
129	Mutations in theSURF1gene associated with Leigh syndrome and cytochromecoxidase deficiency. Human Mutation, 2001, 17, 374-381.	1.1	91
130	RNASEH1 Mutations Impair mtDNA Replication and Cause Adult-Onset Mitochondrial Encephalomyopathy. American Journal of Human Genetics, 2015, 97, 186-193.	2.6	91
131	Microscale oxygraphy reveals OXPHOS impairment in MRC mutant cells. Mitochondrion, 2012, 12, 328-335.	1.6	90
132	Complex IV-deficient Surf1â^'/â^' mice initiate mitochondrial stress responses. Biochemical Journal, 2014, 462, 359-371.	1.7	89
133	Disease-Causing SDHAF1 Mutations Impair Transfer of Fe-S Clusters to SDHB. Cell Metabolism, 2016, 23, 292-302.	7.2	89
134	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.	3.9	89
135	An autosomal locus predisposing to multiple deletions of mtDNA on chromosome 3p. American Journal of Human Genetics, 1996, 58, 763-9.	2.6	88
136	Nuclear DNA origin of cytochrome c oxidase deficiency in Leigh's syndrome: genetic evidence based on patient's-derived rhoÂ transformants. Human Molecular Genetics, 1995, 4, 2017-2023.	1.4	87
137	Early-onset liver mtDNA depletion and late-onset proteinuric nephropathy in Mpv17 knockout mice. Human Molecular Genetics, 2009, 18, 12-26.	1.4	87
138	New genes and pathomechanisms in mitochondrial disorders unraveled by NGS technologies. Biochimica Et Biophysica Acta - Bioenergetics, 2016, 1857, 1326-1335.	0.5	87
139	VARS2andTARS2Mutations in Patients with Mitochondrial Encephalomyopathies. Human Mutation, 2014, 35, 983-989.	1.1	86
140	COQ4 Mutations Cause a Broad Spectrum of Mitochondrial Disorders Associated with CoQ10 Deficiency. American Journal of Human Genetics, 2015, 96, 309-317.	2.6	86
141	MR-1S Interacts with PET100 and PET117 in Module-Based Assembly of Human Cytochrome c Oxidase. Cell Reports, 2017, 18, 1727-1738.	2.9	86
142	Rapamycin rescues mitochondrial myopathy via coordinated activation of autophagy and lysosomal biogenesis. EMBO Molecular Medicine, 2018, 10, .	3.3	86
143	A novel missense adenine nucleotide translocator-1 gene mutation in a Greek adPEO family. Neurology, 2001, 57, 2295-2298.	1.5	85
144	Depletion of mtDNA: Syndromes and genes. Mitochondrion, 2007, 7, 6-12.	1.6	85

#	Article	IF	Citations
145	Respiratory chain and mitochondrial DNA in muscle and brain in Parkinson's disease patients. Neurology, 1993, 43, 2262-2262.	1.5	85
146	Benign reversible muscle cytochrome c oxidase deficiency. Neurology, 1987, 37, 64-64.	1.5	84
147	Chromosomal localization of mitochondrial transcription factor A (TCF6), single-stranded DNA-binding protein (SSBP), and Endonuclease G (ENDOG), three human housekeeping genes involved in mitochondrial biogenesis. Genomics, 1995, 25, 559-564.	1.3	83
148	Mitochondrial Disorders. Medicine (United States), 1998, 77, 59-72.	0.4	83
149	Identification of novel mutations in five patients with mitochondrial encephalomyopathy. Biochimica Et Biophysica Acta - Bioenergetics, 2009, 1787, 491-501.	0.5	83
150	Fatal infantile mitochondrial myopathy and renal dysfunction caused by cytochromec oxidase deficiency: Immunological studies in a new patient. Annals of Neurology, 1985, 17, 414-417.	2.8	82
151	A Third Locus Predisposing to Multiple Deletions of mtDNA in Autosomal Dominant Progressive External Ophthalmoplegia. American Journal of Human Genetics, 1999, 65, 256-260.	2.6	82
152	Decreased aminoacylation of mutant tRNAs in MELAS but not in MERRF patients. Human Molecular Genetics, 2000, 9, 467-475.	1.4	82
153	Nuclear genes in mitochondrial disorders. Current Opinion in Genetics and Development, 2003, 13, 262-270.	1.5	82
154	Myofibrillar-protein isoforms and sarcoplasmic-reticulum Ca2+-transport activity of single human muscle fibres. Biochemical Journal, 1984, 224, 215-225.	1.7	81
155	Dysregulated mitophagy and mitochondrial organization in optic atrophy due to OPA1 mutations. Neurology, 2017, 88, 131-142.	1.5	81
156	Hearing impairment and neurological dysfunction associated with a mutation in the mitochondrial tRNASer(UCN) gene. European Journal of Human Genetics, 1999, 7, 45-51.	1.4	80
157	Genetic and chemical rescue of the Saccharomyces cerevisiae phenotype induced by mitochondrial DNA polymerase mutations associated with progressive external ophthalmoplegia in humans. Human Molecular Genetics, 2006, 15, 2846-2855.	1.4	80
158	Chronic Exposure to Sulfide Causes Accelerated Degradation of Cytochrome c Oxidase in Ethylmalonic Encephalopathy. Antioxidants and Redox Signaling, 2011, 15, 353-362.	2.5	80
159	Mitochondrial disorders. Current Neurology and Neuroscience Reports, 2003, 3, 423-432.	2.0	79
160	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.	4.1	79
161	Fumarase deficiency is an autosomal recessive encephalopathy affecting both the mitochondrial and the cytosolic enzymes. Neurology, 1990, 40, 495-495.	1.5	79
162	Neurological presentations of mitochondrial diseases. Journal of Inherited Metabolic Disease, 1996, 19, 504-520.	1.7	78

#	Article	IF	Citations
163	ETHE1 mutations are specific to ethylmalonic encephalopathy. Journal of Medical Genetics, 2005, 43, 340-346.	1.5	78
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.	1.5	78
165	Loss of apoptosis-inducing factor critically affects MIA40 function. Cell Death and Disease, 2015, 6, e1814-e1814.	2.7	77
166	Myopathy and Fatal Cardiopathy due to Cytochrome c Oxidase Deficiency. Archives of Neurology, 1986, 43, 1198-1202.	4.9	76
167	Genotypes from patients indicate no paternal mitochondrial DNA contribution. Annals of Neurology, 2003, 54, 521-524.	2.8	76
168	Human diseases associated with defects in assembly of OXPHOS complexes. Essays in Biochemistry, 2018, 62, 271-286.	2.1	75
169	Constitutive knockout of Surf1 is associated with high embryonic lethality, mitochondrial disease and cytochrome c oxidase deficiency in mice. Human Molecular Genetics, 2003, 12, 399-413.	1.4	74
170	Effects of riboflavin in children with complex II deficiency. Brain and Development, 2006, 28, 576-581.	0.6	74
171	PINK1heterozygous rare variants: prevalence, significance and phenotypic spectrum. Human Mutation, 2008, 29, 565-565.	1.1	74
172	Carnitine in muscle, serum, and urine of nonprofessional athletes: Effects of physical exercise, training, and L-carnitine administration. Muscle and Nerve, 1991, 14, 598-604.	1.0	73
173	Rare Primary Mitochondrial DNA Mutations and Probable Synergistic Variants in Leberâ€™s Hereditary Optic Neuropathy. PLoS ONE, 2012, 7, e42242.	1.1	73
174	Effective AAVâ€mediated gene therapy in a mouse model of ethylmalonic encephalopathy. EMBO Molecular Medicine, 2012, 4, 1008-1014.	3.3	72
175	Mitochondrial Structure and Bioenergetics in Normal and Disease Conditions. International Journal of Molecular Sciences, 2021, 22, 586.	1.8	72
176	Mitochondrial myopathies. Journal of Inherited Metabolic Disease, 1987, 10, 113-128.	1.7	71
177	Mutations in AAC2, equivalent to human adPEO-associated ANT1 mutations, lead to defective oxidative phosphorylation in Saccharomyces cerevisiae and affect mitochondrial DNA stability. Human Molecular Genetics, 2004, 13, 923-934.	1.4	71
178	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.	0.5	71
179	Paroxysmal non-kinesigenic dyskinesia is caused by mutations of the MR-1 mitochondrial targeting sequence. Human Molecular Genetics, 2009, 18, 1058-1064.	1.4	70
180	Localized cerebral energy failure in DNA polymerase gamma-associated encephalopathy syndromes. Brain, 2010, 133, 1428-1437.	3.7	70

#	Article	IF	Citations
181	Mitochondrial complex III Rieske Fe-S protein processing and assembly. Cell Cycle, 2018, 17, 681-687.	1.3	70
182	Perturbed Redox Signaling Exacerbates a Mitochondrial Myopathy. Cell Metabolism, 2018, 28, 764-775.e5.	7.2	70
183	Biochemical-Clinical Correlation in Patients With Different Loads of the Mitochondrial DNA T8993G Mutation. Archives of Neurology, 2002, 59, 264.	4.9	69
184	Molecular insight into mitochondrial DNA depletion syndrome in two patients with novel mutations in the deoxyguanosine kinase and thymidine kinase 2 genes. Molecular Genetics and Metabolism, 2005, 84, 75-82.	0.5	69
185	Sym1, the yeast ortholog of the MPV17 human disease protein, is a stress-induced bioenergetic and morphogenetic mitochondrial modulator. Human Molecular Genetics, 2010, 19, 1098-1107.	1.4	69
186	MR findings in Leigh syndrome with COX deficiency and SURF-1 mutations. American Journal of Neuroradiology, 2002, 23, 1095-100.	1.2	69
187	A missense mutation in the mitochondrial ND5 gene associated with a Leigh-MELAS overlap syndrome. Neurology, 2003, 60, 1857-1861.	1.5	68
188	Hepatocerebral Form of Mitochondrial DNA Depletion Syndrome. Archives of Neurology, 2008, 65, 1108-13.	4.9	68
189	Redefining phenotypes associated with mitochondrial DNA single deletion. Journal of Neurology, 2015, 262, 1301-1309.	1.8	68
190	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.	1.1	67
191	Defective respiratory capacity and mitochondrial protein synthesis in transformant cybrids harboring the tRNA(Leu(UUR)) mutation associated with maternally inherited myopathy and cardiomyopathy.. Journal of Clinical Investigation, 1994, 93, 1102-1107.	3.9	67
192	Fulminant Leigh syndrome and sudden unexpected death in a family with the T9176C mutation of the mitochondrial ATPase 6 gene. Journal of Inherited Metabolic Disease, 1998, 21, 2-8.	1.7	66
193	Novel mutations in COX15 in a long surviving Leigh syndrome patient with cytochrome c oxidase deficiency. Journal of Medical Genetics, 2005, 42, e28-e28.	1.5	66
194	155th ENMC workshop: Polymerase gamma and disorders of mitochondrial DNA synthesis, 21â€“23 September 2007, Naarden, The Netherlands. Neuromuscular Disorders, 2008, 18, 259-267.	0.3	65
195	Reversible infantile respiratory chain deficiency is a unique, genetically heterogenous mitochondrial disease. Journal of Medical Genetics, 2011, 48, 660-668.	1.5	65
196	Liver transplant in ethylmalonic encephalopathy: a new treatment for an otherwise fatal disease. Brain, 2016, 139, 1045-1051.	3.7	65
197	Disorders from perturbations of nuclearâ€mitochondrial intergenomic crossâ€talk. Journal of Internal Medicine, 2009, 265, 174-192.	2.7	64
198	Mutations in APOPT1, Encoding a Mitochondrial Protein, Cause Cavitating Leukoencephalopathy with Cytochrome c Oxidase Deficiency. American Journal of Human Genetics, 2014, 95, 315-325.	2.6	64

#	Article	IF	Citations
199	TTC19 Plays a Husbandry Role on UQCRFS1 Turnover in the Biogenesis of Mitochondrial Respiratory Complex III. Molecular Cell, 2017, 67, 96-105.e4.	4.5	64
200	Shy1p occurs in a high molecular weight complex and is required for efficient assembly of cytochrome c oxidase in yeast. FEBS Letters, 2001, 498, 46-51.	1.3	63
201	Defective mitochondrial rRNA methyltransferase MRM2 causes MELAS-like clinical syndrome. Human Molecular Genetics, 2017, 26, 4257-4266.	1.4	63
202	Cardiomyopathies in disorders of oxidative metabolism. Cardiovascular Research, 1997, 35, 184-199.	1.8	62
203	Idebenone treatment in patients with OPA1-mutant dominant optic atrophy. Brain, 2013, 136, e231-e231.	3.7	62
204	A Single Cell Complementation Class is Common to Several Cases of Cytochrome c Oxidase-Defective Leigh's Syndrome. Human Molecular Genetics, 1997, 6, 221-228.	1.4	61
205	Disorders of Nuclear-Mitochondrial Intergenomic Communication. Bioscience Reports, 2007, 27, 39-51.	1.1	61
206	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. Orphanet Journal of Rare Diseases, 2018, 13, 120.	1.2	61
207	Early-onset encephalomyopathy associated with tissue-specific mitochondrial DNA depletion: A morphological, biochemical and molecular-genetic study. Journal of Neurology, 1995, 242, 547-556.	1.8	60
208	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.	1.1	60
209	Defective <scp>PITRM</scp> 1 mitochondrial peptidase is associated with AÎ² amyloidotic neurodegeneration. EMBO Molecular Medicine, 2016, 8, 176-190.	3.3	60
210	Cytochrome Oxidase Deficiency: Clinical and Biochemical Heterogeneity. Annals of the New York Academy of Sciences, 1986, 488, 19-32.	1.8	60
211	Phenylbutyrate Therapy for Pyruvate Dehydrogenase Complex Deficiency and Lactic Acidosis. Science Translational Medicine, 2013, 5, 175ra31.	5.8	59
212	Inhibition of proteasome rescues a pathogenic variant of respiratory chain assembly factor COA7. EMBO Molecular Medicine, 2019, 11, .	3.3	59
213	Mitochondrial Diseases. Neurologic Clinics, 1989, 7, 123-156.	0.8	58
214	The polymorphic polyglutamine repeat in the mitochondrial DNA polymerase Î³ gene is not associated with oligozoospermia. Journal of Endocrinological Investigation, 2006, 29, 1-4.	1.8	58
215	International Workshop:. Neuromuscular Disorders, 2017, 27, 1126-1137.	0.3	58
216	miRâ€181a/b downregulation exerts a protective action on mitochondrial disease models. EMBO Molecular Medicine, 2019, 11, .	3.3	58

#	Article	IF	Citations
217	RNase H1 directs origin-specific initiation of DNA replication in human mitochondria. PLoS Genetics, 2019, 15, e1007781.	1.5	58
218	Cytochrome c oxidase deficiency. Biochemical Society Transactions, 1985, 13, 651-653.	1.6	56
219	OPA1 Mutations Associated with Dominant Optic Atrophy Influence Optic Nerve Head Size. Ophthalmology, 2010, 117, 1547-1553.	2.5	56
220	Early Macular Retinal Ganglion Cell Loss in Dominant Optic Atrophy: Genotype-Phenotype Correlation. American Journal of Ophthalmology, 2014, 158, 628-636.e3.	1.7	56
221	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.	2.6	56
222	Impaired Mitochondrial ATP Production Downregulates Wnt Signaling via ER Stress Induction. Cell Reports, 2019, 28, 1949-1960.e6.	2.9	56
223	Identification of new mutations in the ETHE1 gene in a cohort of 14 patients presenting with ethylmalonic encephalopathy. Journal of Medical Genetics, 2008, 45, 473-478.	1.5	55
224	EFNS guidelines on the molecular diagnosis of mitochondrial disorders. European Journal of Neurology, 2009, 16, 1255-1264.	1.7	55
225	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.	3.7	55
226	Sequence of cDNAs encoding subunit Vb of human and bovine cytochrome c oxidase. Gene, 1988, 65, 1-11.	1.0	54
227	Nucleus-driven mutations of human mitochondrial DNA. Journal of Inherited Metabolic Disease, 1992, 15, 456-471.	1.7	54
228	Mitochondrial disorders. Molecular Human Reproduction, 1997, 3, 133-148.	1.3	54
229	Depletion of mitochondrial DNA in fibroblast cultures from patients with POLG1 mutations is a consequence of catalytic mutations. Human Molecular Genetics, 2008, 17, 2496-2506.	1.4	54
230	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.	3.3	54
231	Mitochondrial Myopathy due to Complex III Deficiency With Normal Reducible Cytochrome b Concentration. Archives of Neurology, 1986, 43, 957-961.	4.9	53
232	SURF1 deficiency causes demyelinating Charcot-Marie-Tooth disease. Neurology, 2013, 81, 1523-1530.	1.5	53
233	Mitochondrial DNA sequence characteristics modulate the size of the genetic bottleneck. Human Molecular Genetics, 2016, 25, 1031-1041.	1.4	53
234	Cytochrome c oxidase deficiency. Biochimica Et Biophysica Acta - Bioenergetics, 2021, 1862, 148335.	0.5	53

#	Article	IF	Citations
235	Liver mtDNA content increases during development: A comparison of methods and the importance of age- and tissue-specific controls for the diagnosis of mtDNA depletion. Mitochondrion, 2007, 7, 386-395.	1.6	51
236	EFNS guidelines on the molecular diagnosis of neurogenetic disorders: general issues, Huntingtonâ€™s disease, Parkinsonâ€™s disease and dystonias. European Journal of Neurology, 2009, 16, 777-785.	1.7	51
237	Evolution Meets Disease: Penetrance and Functional Epistasis of Mitochondrial tRNA Mutations. PLoS Genetics, 2011, 7, e1001379.	1.5	51
238	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.	5.0	51
239	Defects in maintenance of mitochondrial DNA are associated with intramitochondrial nucleotide imbalances. Human Molecular Genetics, 2007, 16, 1400-1411.	1.4	50
240	AAV9-based gene therapy partially ameliorates the clinical phenotype of a mouse model of Leigh syndrome. Gene Therapy, 2017, 24, 661-667.	2.3	50
241	Order of Six Loci at 2q24-q31 and Orientation of the HOXD Locus. Genomics, 1994, 24, 34-40.	1.3	49
242	Two novel POLG1 mutations in a patient with progressive external ophthalmoplegia, levodopa-responsive pseudo-orthostatic tremor and parkinsonism. Neuromuscular Disorders, 2008, 18, 460-464.	0.3	49
243	Epstein-Barr Virus Immediate-Early Protein Zta Co-Opts Mitochondrial Single-Stranded DNA Binding Protein To Promote Viral and Inhibit Mitochondrial DNA Replication. Journal of Virology, 2008, 82, 4647-4655.	1.5	49
244	Singleâ€cell expression profiling of dopaminergic neurons combined with association analysis identifies pyridoxal kinase as Parkinson's disease gene. Annals of Neurology, 2009, 66, 792-798.	2.8	49
245	EFNS guidelines on the molecular diagnosis of ataxias and spastic paraplegias. European Journal of Neurology, 2010, 17, 179-188.	1.7	49
246	Cavitating leukoencephalopathy with multiple mitochondrial dysfunction syndrome and NFU1 mutations. Frontiers in Genetics, 2014, 5, 412.	1.1	49
247	Subunit Va of human and bovine cytochrome c oxidase is highly conserved. Gene, 1988, 69, 245-256.	1.0	47
248	Superoxide dismutase gene mutations in Italian patients with familial and sporadic amyotrophic lateral sclerosis: identification of three novel missense mutations. Neuromuscular Disorders, 2001, 11, 404-410.	0.3	47
249	Novel heteroplasmic mtDNA mutation in a family with heterogeneous clinical presentations. Annals of Neurology, 2002, 51, 118-122.	2.8	47
250	AAV-mediated Liver-specific MPV17 Expression Restores mtDNA Levels and Prevents Diet-induced Liver Failure. Molecular Therapy, 2014, 22, 10-17.	3.7	47
251	Strategies for fighting mitochondrial diseases. Journal of Internal Medicine, 2020, 287, 665-684.	2.7	47
252	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.	1.7	47

#	Article	IF	Citations
253	McArdle's disease: Biochemical and molecular genetic studies. Annals of Neurology, 1988, 24, 774-781.	2.8	46
254	POLG1 in idiopathic Parkinson disease. Neurology, 2006, 67, 1698-1700.	1.5	46
255	Molecular defects in cytochrome oxidase in mitochondrial diseases. Journal of Bioenergetics and Biomembranes, 1988, 20, 353-364.	1.0	45
256	Molecular Phenotype of the np 7472 Deafness-Associated Mitochondrial Mutation in Osteosarcoma Cell Cybrids. Human Molecular Genetics, 1999, 8, 2275-2283.	1.4	44
257	Mitochondrial disorders. Current Opinion in Neurology, 2003, 16, 585-594.	1.8	44
258	Glucose metabolism and diet-based prevention of liver dysfunction in MPV17 mutant patients. Journal of Hepatology, 2009, 50, 215-221.	1.8	44
259	Recessive mutations in MSTO1 cause mitochondrial dynamics impairment, leading to myopathy and ataxia. Human Mutation, 2017, 38, 970-977.	1.1	44
260	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.	3.3	43
261	A novel mutation in the mitochondrial tRNAVal gene associated with a complex neurological presentation. Annals of Neurology, 1998, 43, 98-101.	2.8	42
262	Deletions of mitochondrial DNA in Kearns-Sayre syndrome. Neurology, 1998, 51, 1525.	1.5	42
263	Post-transcriptional Silencing and Functional Characterization of the Drosophila melanogaster Homolog of Human Surf1. Genetics, 2006, 172, 229-241.	1.2	42
264	A double mutation (A8296G and G8363A) in the mitochondrial DNA tRNA ^{Lys} gene associated with myoclonus epilepsy with ragged-red fibers. Neurology, 1999, 52, 377-377.	1.5	42
265	Collated mutations in mitochondrial DNA (mtDNA) depletion syndrome (excluding the mitochondrial) Tj ETQq1 1 0.784314 rgBT /Ove 1109-1112.	1.8	41
266	Defects of Mitochondrial DNA. Brain Pathology, 1992, 2, 121-132.	2.1	40
267	COA7(C1orf163/RESA1) mutations associated with mitochondrial leukoencephalopathy and cytochrome c oxidase deficiency. Journal of Medical Genetics, 2016, 53, 846-849.	1.5	40
268	Clinicopathological and genetic studies of two further Italian families with cerebral autosomal dominant arteriopathy. Acta Neuropathologica, 1996, 92, 115-122.	3.9	39
269	Variable penetrance of a familial progressive necrotising encephalopathy due to a novel tRNAIle homoplasmic mutation in the mitochondrial genome. Journal of Medical Genetics, 2004, 41, 342-349.	1.5	39
270	OPA1 mutations and mitochondrial DNA damage: keeping the magic circle in shape. Brain, 2008, 131, 314-317.	3.7	39

#	Article	IF	Citations
271	Altered Sulfide (H2S) Metabolism in Ethylmalonic Encephalopathy. Cold Spring Harbor Perspectives in Biology, 2013, 5, a011437-a011437.	2.3	39
272	Long-Term Sustained Effect of Liver-Targeted Adeno-Associated Virus Gene Therapy for Mitochondrial Neurogastrointestinal Encephalomyopathy. Human Gene Therapy, 2018, 29, 708-718.	1.4	39
273	Differential expression of genes specifying two isoforms of subunit VIa of human cytochrome c oxidase. Gene, 1992, 119, 307-312.	1.0	38
274	Analysis of the trinucleotide CAG repeat from the human mitochondrial DNA polymerase gene in healthy and diseased individuals. European Journal of Human Genetics, 1999, 7, 140-146.	1.4	38
275	A novel homozygous mutation in SUCLA2 gene identified by exome sequencing. Molecular Genetics and Metabolism, 2012, 107, 403-408.	0.5	38
276	Fatigue and exercise intolerance in mitochondrial diseases. Literature revision and experience of the Italian Network of mitochondrial diseases. Neuromuscular Disorders, 2012, 22, S226-S229.	0.3	38
277	Ataxia in mitochondrial disorders. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2012, 103, 359-372.	1.0	37
278	â€œMitochondrial neuropathiesâ€: A survey from the large cohort of the Italian Network. Neuromuscular Disorders, 2016, 26, 272-276.	0.3	37
279	Cloning and expression of human nebulin cDNAs and assignment of the gene to chromosome 2q31-q32. Genomics, 1988, 2, 249-256.	1.3	36
280	X-inactivation patterns in female Leber's hereditary optic neuropathy patients do not support a strong X-linked determinant. , 1996, 61, 356-362.		36
281	Frequency of DYT1 mutation in early onset primary dystonia in Italian patients. Movement Disorders, 2002, 17, 407-408.	2.2	36
282	Infantile mitochondrial encephalopathy. Seminars in Fetal and Neonatal Medicine, 2011, 16, 205-215.	1.1	36
283	Defective Mitochondrial Adenosine Triphosphate Production in Skeletal Muscle From Patients With Dominant Optic Atrophy Due to OPA1 Mutations. Archives of Neurology, 2011, 68, 67-73.	4.9	36
284	Separation of intact pyruvate dehydrogenase complex using blue native agarose gel electrophoresis. Electrophoresis, 2000, 21, 2925-2931.	1.3	35
285	Morphologic evidence of diffuse vascular damage in human and in the experimental model of ethylmalonic encephalopathy. Journal of Inherited Metabolic Disease, 2012, 35, 451-458.	1.7	35
286	Decreased in vitro Mitochondrial Function is Associated with Enhanced Brain Metabolism, Blood Flow, and Memory in Surf1-Deficient Mice. Journal of Cerebral Blood Flow and Metabolism, 2013, 33, 1605-1611.	2.4	35
287	A full-length cDNA encoding a mitochondrial DNA-specific single-stranded DNA binding protein fromXenopus laevis. Nucleic Acids Research, 1991, 19, 4291-4291.	6.5	34
288	Metabolic myopathies. American Journal of Medical Genetics Part A, 1986, 25, 635-651.	2.4	33

#	Article	IF	Citations
289	OXPHOS defects and mitochondrial DNA mutations in cardiomyopathy. Muscle and Nerve, 1995, 18, S170-S174.	1.0	33
290	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.	3.3	33
291	Myoclonus in mitochondrial disorders. Movement Disorders, 2014, 29, 722-728.	2.2	33
292	A novel mutation (8342Gât'A) in the mitochondrial tRNALys gene associated with progressive external ophthalmoplegia and myoclonus. Neuromuscular Disorders, 1999, 9, 66-71.	0.3	32
293	Ultrastructural Analysis of Extraocular Muscle in Chronic Progressive External Ophthalmoplegia. JAMA Ophthalmology, 2000, 118, 1441.	2.6	32
294	Additive effects of POLG1 and ANT1 mutations in a complex encephalomyopathy. Neuromuscular Disorders, 2008, 18, 465-470.	0.3	32
295	Revisiting mitochondrial ocular myopathies: a study from the Italian Network. Journal of Neurology, 2017, 264, 1777-1784.	1.8	32
296	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.	1.8	32
297	A Novel Mutation in the SURF1 Gene in a Child With Leigh Disease, Peripheral Neuropathy, and Cytochrome-c Oxidase Deficiency. Journal of Child Neurology, 2002, 17, 233-236.	0.7	31
298	Proteome adaptations in Ethe1-deficient mice indicate a role in lipid catabolism and cytoskeleton organization via post-translational protein modifications. Bioscience Reports, 2013, 33, .	1.1	31
299	UCP4C mediates uncoupled respiration in larvae of Drosophila melanogaster. EMBO Reports, 2014, 15, 586-591.	2.0	31
300	ATPase Domain <scp>AFG3L2</scp> Mutations Alter <scp>OPA1</scp> Processing and Cause Optic Neuropathy. Annals of Neurology, 2020, 88, 18-32.	2.8	31
301	How Do Human Cells React to the Absence of Mitochondrial DNA?. PLoS ONE, 2009, 4, e5713.	1.1	31
302	The mitochondrial DNA C3303T mutation can cause cardiomyopathy and/or skeletal myopathy. Journal of Pediatrics, 1999, 135, 197-202.	0.9	30
303	Characterization and Expression of the MouseEndonuclease GGene. DNA and Cell Biology, 1997, 16, 1111-1122.	0.9	29
304	Instability of mitochondrial DNA and MRI and clinical correlations in malignant gliomas. Journal of Neuro-Oncology, 2005, 74, 87-90.	1.4	29
305	EFNS guidelines for the molecular diagnosis of neurogenetic disorders: motoneuron, peripheral nerve and muscle disorders. European Journal of Neurology, 2011, 18, 207-217.	1.7	29
306	Improved insulin sensitivity associated with reduced mitochondrial complex IV assembly and activity. FASEB Journal, 2013, 27, 1371-1380.	0.2	29

#	Article	IF	Citations
307	A homozygous MRPL24 mutation causes a complex movement disorder and affects the mitoribosome assembly. Neurobiology of Disease, 2020, 141, 104880.	2.1	29
308	Is nebulin the defective gene product in Duchenne muscular dystrophy?. New England Journal of Medicine, 1987, 316, 107-8.	13.9	29
309	Mitochondrial Neurodegeneration. Cells, 2022, 11, 637.	1.8	29
310	Mitochondrial Retinopathies. International Journal of Molecular Sciences, 2022, 23, 210.	1.8	29
311	Frequency and phenotypes of LRRK2 G2019S mutation in Italian patients with Parkinson's disease. Movement Disorders, 2006, 21, 1232-1235.	2.2	28
312	A novel mutation in the mitochondrial tRNAPro gene associated with late-onset ataxia, retinitis pigmentosa, deafness, leukoencephalopathy and complex I deficiency. European Journal of Human Genetics, 2009, 17, 1092-1096.	1.4	28
313	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.	1.5	28
314	Mitochondria1 encephalomyopathy and partial cytochrome c oxidase deficiency. Neurology, 1987, 37, 58-58.	1.5	28
315	Demonstration and characterization of anti-human mitochondria autoantibodies in idiopathic hypoparathyroidism and in other conditions. Clinical and Experimental Immunology, 1985, 62, 353-60.	1.1	28
316	Oxidative phosphorylation dysfunction does not increase the rate of accumulation of age-related mtDNA deletions in skeletal muscle. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 1997, 379, 1-11.	0.4	27
317	GTP-cyclohydrolase I gene mutations in patients with autosomal dominant and recessive GTP-CH1 deficiency: Identification and functional characterization of four novel mutations. Journal of Inherited Metabolic Disease, 2004, 27, 455-463.	1.7	27
318	Mitochondrial Diseases: A Cross-Talk Between Mitochondrial and Nuclear Genomes. Advances in Experimental Medicine and Biology, 2009, 652, 69-84.	0.8	27
319	Effects of thyroid hormones on the biochemical specialization of human muscle fibers. Muscle and Nerve, 1985, 8, 363-371.	1.0	26
320	Cytochrome Oxidase Deficiency: Clinical and Biochemical Heterogeneity. Annals of the New York Academy of Sciences, 1986, 488, 19-32.	1.8	26
321	Cohen syndrome resulting from a novel large intragenic COH1 deletion segregating in an isolated Greek island population. American Journal of Medical Genetics, Part A, 2008, 146A, 2221-2226.	0.7	26
322	Mitochondrial dementia: A sporadic case of progressive cognitive and behavioral decline with hearing loss due to the rare m.3291T>C MELAS mutation. Journal of the Neurological Sciences, 2011, 300, 165-168.	0.3	26
323	Mitochondrial PITRM1 peptidase loss-of-function in childhood cerebellar atrophy. Journal of Medical Genetics, 2018, 55, 599-606.	1.5	26
324	Epilepsia partialis continua associated with NADH-coenzyme Q reductase deficiency. Journal of the Neurological Sciences, 1995, 129, 152-161.	0.3	25

#	Article	IF	Citations
325	New Splicing-site Mutations in the SURF1Gene in Leigh Syndrome Patients. Journal of Biological Chemistry, 2001, 276, 15326-15329.	1.6	25
326	Monomelic amyotrophy associated with the 7472insC mutation in the mtDNA tRNASer(UCN) gene. Neuromuscular Disorders, 2004, 14, 723-726.	0.3	25
327	A novel de novo dominant mutation inISCUassociated with mitochondrial myopathy. Journal of Medical Genetics, 2017, 54, 815-824.	1.5	25
328	A two-nuclease pathway involving RNase H1 is required for primer removal at human mitochondrial OriL. Nucleic Acids Research, 2018, 46, 9471-9483.	6.5	25
329	Leigh syndrome transmitted by uniparental disomy of chromosome 9. Journal of Medical Genetics, 1999, 36, 927-8.	1.5	25
330	The expanding spectrum of nuclear gene mutations in mitochondrial disorders. Seminars in Cell and Developmental Biology, 2001, 12, 407-416.	2.3	24
331	A nonsense mutation of human <scp>XRCC</scp> 4 is associated with adultâ€onset progressive encephalocardiomyopathy. EMBO Molecular Medicine, 2015, 7, 918-929.	3.3	24
332	SURF1 knockout cloned pigs: Early onset of a severe lethal phenotype. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 2131-2142.	1.8	24
333	Mitochondrial DNA deletion in a girl with manifestations of Kearns-Sayre and Lowe syndromes: An example of phenotypic mimicry?. American Journal of Medical Genetics Part A, 1991, 41, 301-305.	2.4	23
334	EFNS task force on molecular diagnosis of neurologic disorders Guidelines for the molecular diagnosis of inherited neurologic diseases First of two parts. European Journal of Neurology, 2001, 8, 299-314.	1.7	23
335	Predicting the contribution of novel POLG mutations to human disease through analysis in yeast model. Mitochondrion, 2011, 11, 182-190.	1.6	23
336	Common and Novel TMEM70 Mutations in a Cohort of Italian Patients with Mitochondrial Encephalocardiomyopathy. JIMD Reports, 2014, 15, 71-8.	0.7	23
337	Mutations in TIMM50 compromise cell survival in OxPhosâ€dependent metabolic conditions. EMBO Molecular Medicine, 2018, 10, .	3.3	23
338	Searching for genes affecting the structural integrity of the mitochondrial genome. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 1995, 1271, 153-158.	1.8	22
339	Bilateral Putaminal Necrosis Associated With the Mitochondrial DNA A8344G Myoclonus Epilepsy With Ragged Red Fibers (MERRF) Mutation: An Infantile Case. Journal of Child Neurology, 2006, 21, 79-82.	0.7	22
340	Leigh Syndrome in Drosophila melanogaster. Journal of Biological Chemistry, 2014, 289, 29235-29246.	1.6	22
341	Myoclonus epilepsy in mitochondrial disorders. Epileptic Disorders, 2016, 18, 94-102.	0.7	22
342	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.	1.8	22

#	Article	IF	Citations
343	MRI in Leigh syndrome with SURF1 gene mutation. Annals of Neurology, 2002, 51, 138-149.	2.8	21
344	Sequence analysis of familial PEO shows additional mutations associated with the 752C?T and 3527C?T changes in thePOLG1 gene. Annals of Neurology, 2004, 56, 454-455.	2.8	21
345	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.	1.2	21
346	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.	1.8	21
347	Exome sequencing coupled with mRNA analysis identifies NDUFAF6 as a Leigh gene. Molecular Genetics and Metabolism, 2016, 119, 214-222.	0.5	21
348	Expanding the molecular and phenotypic spectrum of truncating MT-ATP6 mutations. Neurology: Genetics, 2020, 6, e381.	0.9	21
349	Loss of COX4I1 Leads to Combined Respiratory Chain Deficiency and Impaired Mitochondrial Protein Synthesis. Cells, 2021, 10, 369.	1.8	21
350	X-Inactivation patterns in females harboring mtDNA mutations that cause Leber hereditary optic neuropathy. Molecular Vision, 2007, 13, 2339-43.	1.1	21
351	Progressive myoclonus epilepsies: an electroclinical, biochemical, morphological and molecular genetic study of 17 cases. Acta Neurologica Scandinavica, 1993, 87, 219-223.	1.0	20
352	Zidovudine Administration during Pregnancy and Mitochondrial Disease in the offspring. Antiviral Therapy, 2005, 10, 697-699.	0.6	20
353	A Novel Insertion Mutation (A169i) in the CLN1 Gene Is Associated with Infantile Neuronal Ceroid Lipofuscinosis in an Italian Patient. Biochemical and Biophysical Research Communications, 1998, 245, 519-522.	1.0	19
354	Multiple origins of the mtDNA 7472insC mutation associated with hearing loss and neurological dysfunction. European Journal of Human Genetics, 2001, 9, 385-387.	1.4	19
355	A Novel Homozygous YARS2 Mutation in Two Italian Siblings and a Review of Literature. JIMD Reports, 2014, 20, 95-101.	0.7	19
356	Clinicopathologic and molecular spectrum of RNASEH1 -related mitochondrial disease. Neurology: Genetics, 2017, 3, e149.	0.9	19
357	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.	1.3	19
358	Lipomatosis Incidence and Characteristics in an Italian Cohort of Mitochondrial Patients. Frontiers in Neurology, 2019, 10, 160.	1.1	19
359	APOPT 1/ COA 8 assists COX assembly and is oppositely regulated by UPS and ROS. EMBO Molecular Medicine, 2019, 11, .	3.3	19
360	Mitochondrial diseases. Neurologic Clinics, 1989, 7, 123-56.	0.8	19

#	Article	IF	Citations
361	Clinical and Biochemical Studies on Cytochrome Oxidase Deficiencies. Annals of the New York Academy of Sciences, 1988, 550, 348-359.	1.8	18
362	The myelin basic protein gene is not a major susceptibility locus for multiple sclerosis in Italian patients. Journal of Neurology, 1994, 241, 615-619.	1.8	18
363	Clinical and EEG findings in eleven patients affected by mitochondrial encephalomyopathy with MERRF-MELAS overlap. Brain and Development, 1996, 18, 185-191.	0.6	18
364	Disorders of mitochondria and related metabolism. Current Opinion in Neurology, 1997, 10, 160-167.	1.8	18
365	Nuclear gene defects in mitochondrial disorders. Neurological Sciences, 1999, 20, 401-408.	0.9	18
366	EFNS task force on molecular diagnosis of neurologic disorders Guidelines for the molecular diagnosis of inherited neurologic diseases Second of two parts. European Journal of Neurology, 2001, 8, 407-424.	1.7	18
367	Identical large scale rearrangement of mitochondrial DNA causes Kearns-Sayre syndrome in a mother and her son. Journal of Medical Genetics, 2003, 40, 858-863.	1.5	18
368	Train, train, train! No pain, just gain. Brain, 2008, 131, 2809-2811.	3.7	18
369	The R336Q mutation in human mitochondrial EFTu prevents the formation of an active mt-EFTuÂ·GTPÂ·aa-tRNA ternary complex. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2009, 1792, 791-795.	1.8	18
370	Hepatocerebral form of mitochondrial DNA depletion syndrome due to mutation in MPV17 gene. Saudi Journal of Gastroenterology, 2012, 18, 285.	0.5	18
371	MELAS-like encephalomyopathy caused by a new pathogenic mutation in the mitochondrial DNA encoded cytochrome c oxidase subunit I. Neuromuscular Disorders, 2012, 22, 990-994.	0.3	18
372	RCC1L (WBSCR16) isoforms coordinate mitochondrial ribosome assembly through their interaction with GTPases. PLoS Genetics, 2020, 16, e1008923.	1.5	18
373	Evidence of Linkage between Susceptibility to Multiple Sclerosis and HLA-Class II Loci in Italian Multiplex Families. European Journal of Human Genetics, 1995, 3, 303-311.	1.4	18
374	Neurological disorders due to mutations of the mitochondrial genome. Neuromuscular Disorders, 1991, 1, 165-172.	0.3	17
375	Mitochondrial myopathy: correlation between oxidative defect and mitochondrial DNA deletions at single fiber level. Acta Neuropathologica, 1994, 87, 371-376.	3.9	17
376	Lack of founder effect for an identical mtDNA depletion syndrome (MDS)-associated MPV17 mutation shared by Navajos and Italians. Neuromuscular Disorders, 2008, 18, 315-318.	0.3	17
377	PGD for the m.14487 T>C mitochondrial DNA mutation resulted in the birth of a healthy boy. Human Reproduction, 2017, 32, 698-703.	0.4	17
378	Role of PITRM1 in Mitochondrial Dysfunction and Neurodegeneration. Biomedicines, 2021, 9, 833.	1.4	17

#	Article	IF	Citations
379	Blue-Native Electrophoresis to Study the OXPHOS Complexes. Methods in Molecular Biology, 2021, 2192, 287-311.	0.4	17
380	Encephalomyopathies caused by abnormal nuclear-mitochondrial intergenomic cross-talk. Acta Myologica, 2009, 28, 2-11.	1.5	17
381	Disorders of nuclear-mitochondrial intergenomic signalling. Journal of Bioenergetics and Biomembranes, 1997, 29, 121-130.	1.0	16
382	NDUFA-1 is not a nuclear modifier gene in Leber hereditary optic neuropathy. Neurology, 2002, 58, 1861-1862.	1.5	16
383	Devic?s neuromyelitis optica and mitochondrial DNA mutation: a case report. Neurological Sciences, 2004, 25, s380-s382.	0.9	16
384	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.	0.7	16
385	Exploiting pyocyanin to treat mitochondrial disease due to respiratory complex III dysfunction. Nature Communications, 2021, 12, 2103.	5.8	16
386	Immunocytochemical study of nebulin in Duchenne muscular dystrophy. Neurology, 1988, 38, 1600-1600.	1.5	15
387	Neuromuscular syndrome associated with the 3291Tâ†'C mutation of mitochondrial DNA: a second case. Neuromuscular Disorders, 2000, 10, 415-418.	0.3	15
388	Mitochondrial disorders. Current Opinion in Neurology, 2001, 14, 553-560.	1.8	15
389	Further pitfalls in the diagnosis of mtDNA mutations: homoplasmic mt-tRNA mutations. Journal of Medical Genetics, 2007, 45, 55-61.	1.5	15
390	A case of Leber hereditary optic neuropathy plus dystonia caused by G14459A mitochondrial mutation. Neurological Sciences, 2013, 34, 407-408.	0.9	15
391	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.	0.7	15
392	Paradoxical Inhibition of Glycolysis by Pioglitazone Opposes the Mitochondriopathy Caused by AIF Deficiency. EBioMedicine, 2017, 17, 75-87.	2.7	15
393	Compound heterozygous missense and deep intronic variants in NDUFAF6 unraveled by exome sequencing and mRNA analysis. Journal of Human Genetics, 2018, 63, 563-568.	1.1	15
394	DNA polymerase gamma mutations that impair holoenzyme stability cause catalytic subunit depletion. Nucleic Acids Research, 2021, 49, 5230-5248.	6.5	15
395	Duplexing complexome profiling with SILAC to study human respiratory chain assembly defects. Biochimica Et Biophysica Acta - Bioenergetics, 2021, 1862, 148395.	0.5	15
396	SILAC-based complexome profiling dissects the structural organization of the human respiratory supercomplexes in SCAFIKO cells. Biochimica Et Biophysica Acta - Bioenergetics, 2021, 1862, 148414.	0.5	15

#	Article	IF	Citations
397	Chapter 30 Mitochondrial disorders. Supplements To Clinical Neurophysiology, 2004, 57, 304-312.	2.1	14
398	The A467T and W748S POLG substitutions are a rare cause of adult-onset ataxia in Europe. Brain, 2006, 130, E69-E69.	3.7	14
399	Clinical and genetic features of paroxysmal kinesigenic dyskinesia in Italian patients. European Journal of Paediatric Neurology, 2016, 20, 152-157.	0.7	14
400	Sequence variations in the NDUFA1 gene encoding a subunit of complex I of the respiratory chain. Journal of Inherited Metabolic Disease, 2001, 24, 15-27.	1.7	13
401	Functional Characterization of drim2, the Drosophila melanogaster Homolog of the Yeast Mitochondrial Deoxynucleotide Transporter. Journal of Biological Chemistry, 2014, 289, 7448-7459.	1.6	13
402	Severe early onset ethylmalonic encephalopathy with West syndrome. Metabolic Brain Disease, 2015, 30, 1537-1545.	1.4	13
403	A family with paroxysmal nonkinesigenic dyskinesias (PNKD): Evidence of mitochondrial dysfunction. European Journal of Paediatric Neurology, 2015, 19, 64-68.	0.7	13
404	Unravelling the Effects of the Mutation m.3571insC/MT-ND1 on Respiratory Complexes Structural Organization. International Journal of Molecular Sciences, 2018, 19, 764.	1.8	13
405	NDUFS3 depletion permits complex I maturation and reveals TMEM126A/OPA7 as an assembly factor binding the ND4-module intermediate. Cell Reports, 2021, 35, 109002.	2.9	13
406	Mitochondrial myopathy and ophthalmoplegia in a sporadic patient with the 5698Gâ†'A mitochondrial DNA mutation. Neuromuscular Disorders, 2004, 14, 815-817.	0.3	12
407	Quantitative proteomics suggests metabolic reprogramming during ETHE1 deficiency. Proteomics, 2016, 16, 1166-1176.	1.3	12
408	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.	1.8	12
409	RNase H1 Regulates Mitochondrial Transcription and Translation via the Degradation of 7S RNA. Frontiers in Genetics, 2019, 10, 1393.	1.1	12
410	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.	2.6	12
411	Hypoxic and hypercapnic challenges unveil respiratory vulnerability of Surf1 knockout mice, an animal model of Leigh syndrome. Mitochondrion, 2011, 11, 413-420.	1.6	11
412	The Gene (NFE2L1) for Human NRF-1, an Activator Involved in Nuclear-Mitochondrial Interactions, Maps to 7q32. Genomics, 1995, 27, 555-557.	1.3	10
413	Utility of multimodal evoked potential study and electroencephalography in mitochondrial encephalomyopathy. Italian Journal of Neurological Sciences, 1998, 19, 291-300.	0.1	10
414	EFNS guidelines on the molecular diagnosis of channelopathies, epilepsies, migraine, stroke, and dementias. European Journal of Neurology, 2010, 17, 641-648.	1.7	10

#	Article	IF	Citations
415	Functional Outcome of Children With Mitochondrial Diseases. Pediatric Neurology, 2011, 44, 340-346.	1.0	10
416	Nonsense mutation in pseudouridylate synthase 1 (PUS1) in two brothers affected by myopathy, lactic acidosis and sideroblastic anaemia (MLASA). BMJ Case Reports, 2009, 2009, bcr0520091889-bcr0520091889.	0.2	10
417	Mitochondrial myopathies. Current Opinion in Rheumatology, 1994, 6, 559-567.	2.0	9
418	Mitochondrial Encephalomyopathy Lactic Acidosis and Strokelike Episodes Mimicking Occipital Idiopathic Epilepsy. Pediatric Neurology, 2009, 41, 131-134.	1.0	9
419	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.	0.5	9
420	Opa1 Overexpression Protects from Early-Onset Mpv17â^'/â^'-Related Mouse Kidney Disease. Molecular Therapy, 2020, 28, 1918-1930.	3.7	9
421	La risonanza magnetica nelle encefalomiopatie mitocondriali. The Neuroradiology Journal, 1992, 5, 25-32.	0.1	8
422	Remarkable Recovery of Visual Function in a Patient with Leber's Optic Neuropathy and Multiple Mutations of Mitochondrial DNA. International Journal of Neuroscience, 1994, 77, 261-266.	0.8	8
423	Mitochondrial Matchmaking. New England Journal of Medicine, 2016, 375, 1894-1896.	13.9	8
424	Pure myopathy with enlarged mitochondria associated to a new mutation in MTND2 gene. Molecular Genetics and Metabolism Reports, 2017, 10, 24-27.	0.4	8
425	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.	0.4	8
426	Effects of pirenzepine on plasma insulin, glucagon and pancreatic polypeptide levels in normal man. European Journal of Clinical Pharmacology, 1985, 27, 701-705.	0.8	7
427	Respiratory-chain and pyruvate metabolism defects: Italian collaborative survey on 72 patients. Journal of Inherited Metabolic Disease, 1996, 19, 143-148.	1.7	7
428	The V368i mutation in Twinkle does not segregate with adPEO. Annals of Neurology, 2003, 53, 278-278.	2.8	7
429	Modelling of BCS1L-related human mitochondrial disease in Drosophila melanogaster. Journal of Molecular Medicine, 2021, 99, 1471-1485.	1.7	7
430	Biochemical and molecular aspects of cytochrome C oxidase deficiency. Advances in Neurology, 1988, 48, 93-105.	0.8	7
431	The relevance of migraine in the clinical spectrum of mitochondrial disorders. Scientific Reports, 2022, 12, 4222.	1.6	7
432	CG7630 is the Drosophila melanogaster homolog of the cytochrome c oxidase subunit COX7B. EMBO Reports, 0, , .	2.0	7

#	Article	IF	Citations
433	Sequence analysis of mitochondrial DNA in a new maternally inherited encephalomyopathy. Journal of Neurology, 1995, 242, 490-496.	1.8	6
434	A Myopathy, Lactic Acidosis, Sideroblastic Anemia (Mlasa) Case Due to A Novel Pus1 Mutation. Turkish Journal of Haematology, 2017, 34, 376-377.	0.2	6
435	Biallelic mutations in NDUFA8 cause complex I deficiency in two siblings with favorable clinical evolution. Molecular Genetics and Metabolism, 2020, 131, 349-357.	0.5	6
436	Mitochondrial diseases. BailliÃ¨re's Clinical Neurology, 1994, 3, 315-34.	0.2	6
437	Severe Infantile Hypotonia With Ethylmalonic Aciduria: Case Report. Journal of Child Neurology, 2008, 23, 703-705.	0.7	5
438	Lactic Acidosis in a Newborn With Adrenal Calcifications. Pediatric Research, 2009, 66, 317-322.	1.1	5
439	LBSL (leukoencephalopathy with brain stem and spinal cord involvement and high lactate) without sparing of the u-fibers and globi pallidi: A case report. European Journal of Radiology Extra, 2011, 79, e73-e76.	0.1	5
440	Adult-onset leukodystrophies from respiratory chain disorders: do they exist?. Journal of Neurology, 2013, 260, 1617-1623.	1.8	5
441	Distributed abnormalities of brain white matter architecture in patients with dominant optic atrophy and OPA1 mutations. Journal of Neurology, 2015, 262, 1216-1227.	1.8	5
442	Myopathic mitochondrial DNA depletion syndrome associated with biallelic variants in LIG3. Brain, 2021, 144, e74-e74.	3.7	5
443	Mitochondrial encephalomyopathies. Progress in Clinical and Biological Research, 1989, 306, 117-28.	0.2	5
444	Mitochondrial Cytochrome c Oxidase Defects Alter Cellular Homeostasis of Transition Metals. Frontiers in Cell and Developmental Biology, 2022, 10, .	1.8	5
445	Variation in MAPT is not a contributing factor to the incomplete penetrance in LHON. Mitochondrion, 2011, 11, 620-622.	1.6	4
446	Breathe: Your Mitochondria Will Do the Restâ€¦ If They Are Healthy!. Cell Metabolism, 2019, 30, 628-629.	7.2	4
447	Neurodevelopmental regression, severe generalized dystonia, and metabolic acidosis caused by POLR3A mutations. Neurology: Genetics, 2020, 6, e521.	0.9	4
448	Genetic variation in the methylenetetrahydrofolate reductase gene, MTHFR, does not alter the risk of visual failure in Leber's hereditary optic neuropathy. Molecular Vision, 2009, 15, 870-5.	1.1	4
449	Variation in OPA1 does not explain the incomplete penetrance of Leber hereditary optic neuropathy. Molecular Vision, 2010, 16, 2760-4.	1.1	4
450	Accelerated cardiomyopathy in maternally inherited diabetes and deafness. International Journal of Clinical Pharmacology Research, 2004, 24, 15-21.	0.4	4

#	Article	IF	Citations
451	The role of APOE in the phenotypic expression of Leber hereditary optic neuropathy. Journal of Medical Genetics, 2003, 40, 41e-41.	1.5	3
452	Dominance in mitochondrial disorders. Journal of Inherited Metabolic Disease, 2005, 28, 287-299.	1.7	3
453	Mitochondrial complex III deficiency in a case of HCV related noninflammatory myopathy. Journal of Neurology, 2007, 254, 1450-1452.	1.8	3
454	Mitochondrial myopathy biomarker Fibroblast growth factor 21 is induced by muscle mtDNA instability and translation defects. Mitochondrion, 2015, 24, S45-S46.	1.6	3
455	Mitochondrial Diseases in Childhood. Current Molecular Medicine, 2014, 14, 1069-1078.	0.6	3
456	Mitochondrial disorders. Brain, 2004, 127, 2783-2783.	3.7	2
457	Partial tandem duplication of mtDNAâ€“tRNAPhe impairs mtDNA translation in late-onset mitochondrial myopathy. Neuromuscular Disorders, 2012, 22, 50-55.	0.3	2
458	Screening for POLG W748S and A467T mutations in ataxia patients from Spain. Movement Disorders, 2012, 27, 1326-1326.	2.2	2
459	Awareness of rare and genetic neurological diseases among italian neurologist. A national survey. Neurological Sciences, 2020, 41, 1567-1570.	0.9	2
460	Mitochondrial Diseases. , 1989, , 157-166.		2
461	Molecular Pathogenesis of Mitochondrial Diseases. Progress in Cell Research, 1995, 5, 223-224.	0.3	2
462	Response to Drs. Djouadi and Bastin. Cell Metabolism, 2011, 14, 717.	7.2	1
463	Data on cytochrome c oxidase assembly in mice and human fibroblasts or tissues induced by SURF1 defect. Data in Brief, 2016, 7, 1004-1009.	0.5	1
464	Mitochondrial Genes and Neurodegenerative Disease. , 2016, , 81-106.		1
465	Metabolic and Mitochondrial Ataxias. , 2003, , 231-252.		1
466	Diagnosis of hereditary cardiomyopathies. Biomedicine and Pharmacotherapy, 1992, 46, 175-176.	2.5	0
467	Pitfalls in restriction fragment length polymorphism analysis of Leber's hereditary optic neuropathy patients. Mitochondrion, 2004, 4, 37-39.	1.6	0
468	Mitochondrial DNA and OXPHOS Disorders. , 2004, , 95-116.		0

#	Article	IF	Citations
469	M.I.3 Nuclear genes involved in mitochondrial syndromes. Neuromuscular Disorders, 2007, 17, 764-765.	0.3	0
470	Depletion of mitochondrial DNA in fibroblast cultures from patients with POLG1 mutations is a consequence of catalytic mutations. Human Molecular Genetics, 2009, 18, 4905-4906.	1.4	0
471	P301 Mitochondrial abnormalities in a newborn with lactic acidosis and adrenal calcifications. European Journal of Paediatric Neurology, 2009, 13, S114-S115.	0.7	0
472	PAW34 Mutations in OPA1 expand the clinical phenotype of mitochondrial disease. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, e32-e33.	0.9	0
473	P55 Reversible infantile respiratory chain deficiency is a genetically heterogenous mitochondrial disease. Neuromuscular Disorders, 2011, 21, S22.	0.3	0
474	Epilepsy in Mitochondrial Disorders. , 2011, , .		0
475	Phenylbutyrate therapy for pyruvate dehydrogenase deficiency. Mitochondrion, 2012, 12, 572-573.	1.6	0
476	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.	3.3	0
477	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.	0.3	0
478	Mitochondrial medicine: Disease genes and mechanisms. Biochimica Et Biophysica Acta - Bioenergetics, 2016, 1857, e6.	0.5	0
479	Recessive mutations in novel gene MST01 cause early onset neuromuscular condition. Neuromuscular Disorders, 2017, 27, S176.	0.3	0
480	MITOCHONDRIAL DISEASES I (Oral). Neuromuscular Disorders, 2018, 28, S87.	0.3	0
481	P12â€…A novel model of cardiomyopathy reveals a tissue specific role for the complex i assembly factor ecsit. , 2018, , .		0
482	Experimental Therapies. , 2019, , 357-370.		0
483	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.	1.4	0
484	300 HEMIZYGOUS PRION PROTEIN GENE (PRNP) KNOCKOUT IN CATTLE FIBROBLASTS. Reproduction, Fertility and Development, 2008, 20, 230.	0.1	0
485	Ethylmalonic Encephalopathy. , 2014, , 157-163.		0
486	Analysis of giant deletions of human mitochondrial DNA in progressive external ophthalmoplegia. , 1991, , 209-220.		0

#	Article	IF	Citations
487	DÃ©lÃ©tions hÃ©ritÃ©es et de novo de la rÃ©gion 5q13 dans les amyotrophies spinales infantiles. Medecine/Sciences, 1994, 10, 889.	0.0	0
488	DerniÃ¨re heure : identification et caractÃ©risation d'un gÃ¨ne dÃ©terminant dans les amyotrophies spinales. Medecine/Sciences, 1995, 11, 149.	0.0	0
489	Nucleus-Driven Lesions of mtDNA and Disorders of Nucleus-Encoded Energy Genes. , 1999, , 729-749.		0
490	Biogenesis of NDUFS3-Less Complex I Indicates TMEM126A/OPA7 as an Assembly Factor of the ND4-Module. SSRN Electronic Journal, 0, , .	0.4	0
491	Mitochondrial myopathy: correlation between oxidative defect and mitochondrial DNA deletions at single fiber level. Acta Neuropathologica, 1994, 87, 371-376.	3.9	0