# Massimo Zeviani

List of Publications by Year
in descending order



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|---|---|---|---|
| 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 | Identification and characterization of a spinal muscular atrophy-determining gene. Cell, 1995, 80, 155-165. | 28.9 | 3,424 |
| 2 | Mitochondrial DNA Deletions in Progressive External Ophthalmoplegia and Kearns-Sayre Syndrome. New England Journal of Medicine, 1989, 320, 1293-1299. | 27.0 | 1,012 |
| 3 | Mitochondrial diseases. Nature Reviews Disease Primers, 2016, 2, 16080. | 30.5 | 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. | 21.4 | 803 |
| 5 | Spastic Paraplegia and OXPHOS Impairment Caused by Mutations in Paraplegin, a Nuclear-Encoded Mitochondrial Metalloprotease. Cell, 1998, 93, 973-983. | 28.9 | 784 |
| 6 | Mitochondrial myopathies. Annals of Neurology, 1985, 17, 521-538. | 5.3 | 762 |
| 7 | Oxygen sensing requires mitochondrial ROS but not oxidative phosphorylation. Cell Metabolism, 2005, 1, 409-414. | 16.2 | 678 |
| 8 | An autosomal dominant disorder with multiple deletions of mitochondrial DNA starting at the D-loop region. Nature, 1989, 339, 309-311. | 27.8 | 640 |
| 9 | Deletions of mitochondrial DNA in Kearnsâ€Sayre syndrome. Neurology, 1988, 38, 1339-1339. | 1.1 | 624 |
| 10 | Role of Adenine Nucleotide Translocator 1 in mtDNA Maintenance. Science, 2000, 289, 782-785. | 12.6 | 591 |
| 11 | A direct repeat is a hotspot for large-scale deletion of human mitochondrial DNA. Science, 1989, 244, 346-349. | 12.6 | 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. | 6.2 | 504 |
| 13 | Mutations of SURF-1 in Leigh Disease Associated with Cytochrome c Oxidase Deficiency. American Journal of Human Genetics, 1998, 63, 1609-1621. | 6.2 | 504 |
| 14 | Deletions of mitochondrial DNA in Kearnsâ€Sayre syndrome. Neurology, 1988, 38, 1339-1339. | 1.1 | 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. | 6.2 | 397 |
| 16 | Phenotypic spectrum associated with mutations of the mitochondrial polymerase Â gene. Brain, 2006, 129, 1674-1684. | 7.6 | 397 |
| 17 | Maternally inherited myopathy and cardiomyopathy: association with mutation in mitochondrial DNA tRNALeu(UUR). Lancet, The, 1991, 338, 143-147. | 13.7 | 395 |
| 18 | Cytochromec oxidase deficiency in leigh syndrome. Annals of Neurology, 1987, 22, 498-506. | 5.3 | 390 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Multi-system neurological disease is common in patients with OPA1 mutations. Brain, 2010, 133, 771-786. | 7.6 | 385 |
| 20 | MPV17 encodes an inner mitochondrial membrane protein and is mutated in infantile hepatic mitochondrial DNA depletion. Nature Genetics, 2006, 38, 570-575. | 21.4 | 380 |
| 21 | Mitochondrial disorders. Brain, 2004, 127, 2153-2172. | 7.6 | 362 |
| 22 | Loss of ETHE1, a mitochondrial dioxygenase, causes fatal sulfide toxicity in ethylmalonic encephalopathy. Nature Medicine, 2009, 15, 200-205. | 30.7 | 358 |
| 23 | The Opa1-Dependent Mitochondrial Cristae Remodeling Pathway Controls Atrophic, Apoptotic, and Ischemic Tissue Damage. Cell Metabolism, 2015, 21, 834-844. | 16.2 | 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. | 7.6 | 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. | 6.2 | 331 |
| 26 | De novo and inherited deletions of the 5q13 region in spinal muscular atrophies. Science, 1994, 264, 1474-1477. | 12.6 | 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. | 6.2 | 326 |
| 28 | Systematic identification of human mitochondrial disease genes through integrative genomics. Nature Genetics, 2006, 38, 576-582. | 21.4 | 321 |
| 29 | Transthyretin. Neurology, 1986, 36, 900-900. | 1.1 | 300 |
| 30 | NAD+-Dependent Activation of Sirt1 Corrects the Phenotype in a Mouse Model of Mitochondrial Disease. Cell Metabolism, 2014, 19, 1042-1049. | 16.2 | 293 |
| 31 | Infantile hepatocerebral syndromes associated with mutations in the mitochondrial DNA polymerase-ÂA. Brain, 2005, 128, 723-731. | 7.6 | 284 |
| 32 | Increased longevity and refractoriness to Ca2+-dependent neurodegeneration in Surf1 knockout mice. Human Molecular Genetics, 2007, 16, 431-444. | 2.9 | 279 |
| 33 | An autosomal locus predisposing to deletions of mitochondrial DNA. Nature Genetics, 1995, 9, 146-151. | 21.4 | 268 |
| 34 | Mutations of mitochondrial DNA polymerase Î³A are a frequent cause of autosomal dominant or recessive progressive external ophthalmoplegia. Annals of Neurology, 2002, 52, 211-219. | 5.3 | 257 |
| 35 | Cytochrome c Oxidase subunit I microdeletion in a patient with motor neuron disease. Annals of Neurology, 1998, 43, 110-116. | 5.3 | 251 |
| 36 | Transcription Factor EB Controls Metabolic Flexibility during Exercise. Cell Metabolism, 2017, 25, 182-196. | 16.2 | 250 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Autosomal recessive mitochondrial ataxic syndrome due to mitochondrial polymerase Î³ mutations. Neurology, 2005, 64, 1204-1208. | 1.1 | 246 |
| 38 | In Vivo Correction of COX Deficiency by Activation of the AMPK/PGC-1Î± Axis. Cell Metabolism, 2011, 14, 80-90. | 16.2 | 245 |
| 39 | Exome sequencing identifies ACAD9 mutations as a cause of complex I deficiency. Nature Genetics, 2010, 42, 1131-1134. | 21.4 | 234 |
| 40 | SDHAF1, encoding a LYR complex-II specific assembly factor, is mutated in SDH-defective infantile leukoencephalopathy. Nature Genetics, 2009, 41, 654-656. | 21.4 | 233 |
| 41 | Clinical and molecular findings in children with complex I deficiency. Biochimica Et Biophysica Acta - Bioenergetics, 2004, 1659, 136-147. | 1.0 | 231 |
| 42 | Efficient mitochondrial biogenesis drives incomplete penetrance in Leberâ€™s hereditary optic neuropathy. Brain, 2014, 137, 335-353. | 7.6 | 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. | 2.9 | 218 |
| 44 | Genome editing in mitochondria corrects a pathogenic mtDNA mutation in vivo. Nature Medicine, 2018, 24, 1691-1695. | 30.7 | 215 |
| 45 | A SIRT7-Dependent Acetylation Switch of GABPÎ²1 Controls Mitochondrial Function. Cell Metabolism, 2014, 20, 856-869. | 16.2 | 214 |
| 46 | Pharmacological Inhibition of Poly(ADP-Ribose) Polymerases Improves Fitness and Mitochondrial Function in Skeletal Muscle. Cell Metabolism, 2014, 19, 1034-1041. | 16.2 | 211 |
| 47 | Mitochondrial medicine: A metabolic perspective on the pathology of oxidative phosphorylation disorders. Cell Metabolism, 2006, 3, 9-13. | 16.2 | 210 |
| 48 | Opa1 Overexpression Ameliorates the Phenotype of Two Mitochondrial Disease Mouse Models. Cell Metabolism, 2015, 21, 845-854. | 16.2 | 202 |
| 49 | Risk of developing a mitochondrial DNA deletion disorder. Lancet, The, 2004, 364, 592-596. | 13.7 | 201 |
| 50 | Severe X-Linked Mitochondrial Encephalomyopathy Associated with a Mutation in Apoptosis-Inducing Factor. American Journal of Human Genetics, 2010, 86, 639-649. | 6.2 | 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. | 2.8 | 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. | 6.2 | 192 |
| 53 | Idebenone Treatment In Leber's Hereditary Optic Neuropathy. Brain, 2011, 134, e188-e188. | 7.6 | 192 |
| 54 | Lack of the Mitochondrial Protein Acylglycerol Kinase Causes Sengers Syndrome. American Journal of Human Genetics, 2012, 90, 314-320. | 6.2 | 192 |

| # | Article | IF | Citations |
|---|---|---|---|
| 55 | Evidence that nebulin is a protein-ruler in muscle thin filaments. FEBS Letters, 1991, 282, 313-316. | 2.8 | 187 |
| 56 | Leukoencephalopathy with thalamus and brainstem involvement and high lactate â€˜LTBLâ€™ caused by EARS2 mutations. Brain, 2012, 135, 1387-1394. | 7.6 | 187 |
| 57 | MELAS syndrome. Neurology, 1988, 38, 751-751. | 1.1 | 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. | 4.1 | 182 |
| 59 | Molecular diagnosis in mitochondrial complex I deficiency using exome sequencing. Journal of Medical Genetics, 2012, 49, 277-283. | 3.2 | 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. | 6.2 | 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. | 2.9 | 178 |
| 62 | POLG1 mutations cause a syndromic epilepsy with occipital lobe predilection. Brain, 2008, 131, 818-828. | 7.6 | 176 |
| 63 | Mutational spectrum of the CHAC gene in patients with chorea-acanthocytosis. European Journal of Human Genetics, 2002, 10, 773-781. | 2.8 | 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. | 6.2 | 172 |
| 65 | Novel (ovario) leukodystrophy related to AARS2 mutations. Neurology, 2014, 82, 2063-2071. | 1.1 | 172 |
| 66 | FGF21 is a biomarker for mitochondrial translation and mtDNA maintenance disorders. Neurology, 2016, 87, 2290-2299. | 1.1 | 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. | 6.2 | 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. | 2.9 | 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. | 6.2 | 165 |
| 70 | Mutations of the Mitochondrial-tRNA Modifier MTO1 Cause Hypertrophic Cardiomyopathy and Lactic Acidosis. American Journal of Human Genetics, 2012, 90, 1079-1087. | 6.2 | 164 |
| 71 | Clinical and molecular features of mitochondrial DNA depletion syndromes. Journal of Inherited Metabolic Disease, 2009, 32, 143-158. | 3.6 | 161 |
| 72 | Impaired complex III assembly associated with BCS1L gene mutations in isolated mitochondrial encephalopathy. Human Molecular Genetics, 2007, 16, 1241-1252. | 2.9 | 158 |

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|---|---|---|---|
| 73 | Isolation of mitochondria for biogenetical studies: An update. Mitochondrion, 2010, 10, 253-262. | 3.4 | 158 |
| 74 | Phenotypic heterogeneity of the 8344A>G mtDNA â€œMERRFâ€•mutation. Neurology, 2013, 80, 2049-2054. | 1.1 | 157 |
| 75 | New treatments for mitochondrial diseaseâ€”no time to drop our standards. Nature Reviews Neurology, 2013, 9, 474-481. | 10.1 | 157 |
| 76 | Expression of the Ciona intestinalis Alternative Oxidase (AOX) in Drosophila Complements Defects in Mitochondrial Oxidative Phosphorylation. Cell Metabolism, 2009, 9, 449-460. | 16.2 | 156 |
| 77 | Syndromic parkinsonism and dementia associated with <scp>OPA</scp>1 missense mutations. Annals of Neurology, 2015, 78, 21-38. | 5.3 | 154 |
| 78 | Fatal infantile cytochrome c oxidase deficiency. Neurology, 1985, 35, 802-802. | 1.1 | 149 |
| 79 | Mutations in TTC19 cause mitochondrial complex III deficiency and neurological impairment in humans and flies. Nature Genetics, 2011, 43, 259-263. | 21.4 | 148 |
| 80 | Mutations of ANT1 , Twinkle , and POLG1 in sporadic progressive external ophthalmoplegia (PEO). Neurology, 2003, 60, 1354-1356. | 1.1 | 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. | 6.2 | 146 |
| 82 | An international classification of inherited metabolic disorders (<scp>ICIMD</scp>). Journal of Inherited Metabolic Disease, 2021, 44, 164-177. | 3.6 | 146 |
| 83 | MtDNAâ€maintenance defects: syndromes and genes. Journal of Inherited Metabolic Disease, 2017, 40, 587-599. | 3.6 | 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. | 2.9 | 144 |
| 85 | Epileptic phenotypes associated with mitochondrial disorders. Neurology, 2001, 56, 1340-1346. | 1.1 | 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. | 2.8 | 143 |
| 87 | Mutations in FBXL4, Encoding a Mitochondrial Protein, Cause Early-Onset Mitochondrial Encephalomyopathy. American Journal of Human Genetics, 2013, 93, 482-495. | 6.2 | 138 |
| 88 | Combined treatment with oral metronidazole and N-acetylcysteine is effective in ethylmalonic encephalopathy. Nature Medicine, 2010, 16, 869-871. | 30.7 | 136 |
| 89 | Hereditary spastic paraplegia is a novel phenotype for GJA12/GJC2 mutations. Brain, 2009, 132, 426-438. | 7.6 | 135 |
| 90 | Characterization of SURF-1 Expression and Surf-1p Function in Normal and Disease Conditions. Human Molecular Genetics, 1999, 8, 2533-2540. | 2.9 | 134 |

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|---|---|---|---|
| 91 | Cowchock Syndrome Is Associated with a Mutation in Apoptosis-Inducing Factor. American Journal of Human Genetics, 2012, 91, 1095-1102. | 6.2 | 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. | 1.9 | 133 |
| 93 | Novel mutations of ND genes in complex I deficiency associated with mitochondrial encephalopathy. Brain, 2007, 130, 1894-1904. | 7.6 | 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. | 3.2 | 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. | 3.4 | 128 |
| 96 | Human mitochondrial complex I assembly is mediated by NDUFAF1. FEBS Journal, 2005, 272, 5317-5326. | 4.7 | 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. | 2.9 | 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. | 8.2 | 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. | 6.2 | 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. | 5.3 | 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. | 2.9 | 120 |
| 102 | Dominantly inherited mitochondrial myopathy with multiple deletions of mitochondrial DNA. Neurology, 1991, 41, 1053-1053. | 1.1 | 120 |
| 103 | The m.3243A>G mitochondrial DNA mutation and related phenotypes. A matter of gender?. Journal of Neurology, 2014, 261, 504-510. | 3.6 | 119 |
| 104 | Complex I deficiency is Associated with 3243G:C Mitochondrial DNA in Osteosarcoma Cell Cybrids. Human Molecular Genetics, 1996, 5, 123-129. | 2.9 | 117 |
| 105 | Mitochondrial disorders of the OXPHOS system. FEBS Letters, 2021, 595, 1062-1106. | 2.8 | 117 |
| 106 | A novel mtDNA mutation in the ND5 subunit of complex I in two MELAS patients. Annals of Neurology, 2001, 49, 106-110. | 5.3 | 116 |
| 107 | Assembly Factors of Human Mitochondrial Respiratory Chain Complexes: Physiology and Pathophysiology. Advances in Experimental Medicine and Biology, 2012, 748, 65-106. | 1.6 | 116 |
| 108 | Nuclear gene mutations as the cause of mitochondrial complex III deficiency. Frontiers in Genetics, 2015, 6, 134. | 2.3 | 116 |

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|---|---|---|---|
| 109 | Genotype to phenotype correlations in mitochondrial encephalomyopathies associated with the A3243G mutation of mitochondrial DNA. Journal of Neurology, 1995, 242, 304-312. | 3.6 | 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. | 6.2 | 115 |
| 111 | Myoclonic epilepsy and ragged-red fibers with cytochrome oxidase deficiency: Neuropathology, biochemistry, and molecular genetics. Annals of Neurology, 1989, 26, 20-33. | 5.3 | 111 |
| 112 | Mitochondrial DNA mutations in patients with postlingual, nonsyndromic hearing impairment. European Journal of Human Genetics, 2005, 13, 26-33. | 2.8 | 110 |
| 113 | Isolation of a cDNA clone encoding subunit IV of human cytochrome c oxidase. Gene, 1987, 55, 205-217. | 2.2 | 109 |
| 114 | Cloning of human and rat cDNAs encoding the mitochondrial single-stranded DNA-binding protein (SSB). Gene, 1993, 126, 219-225. | 2.2 | 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. | 2.9 | 109 |
| 116 | Disorders of nuclear-mitochondrial intergenomic signaling. Gene, 2005, 354, 162-168. | 2.2 | 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. | 6.2 | 108 |
| 118 | Mitochondrial disorders. Current Opinion in Neurology, 2007, 20, 564-571. | 3.6 | 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. | 6.2 | 107 |
| 120 | Mutations in COX7B Cause Microphthalmia with Linear Skin Lesions, an Unconventional Mitochondrial Disease. American Journal of Human Genetics, 2012, 91, 942-949. | 6.2 | 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. | 7.8 | 102 |
| 122 | Peripheral neuropathy in mitochondrial disorders. Lancet Neurology, The, 2013, 12, 1011-1024. | 10.2 | 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. | 0.6 | 100 |
| 124 | Tissue distribution and transmission of mitochondrial DNA deletions in mitochondrial myopathies. Annals of Neurology, 1990, 28, 94-97. | 5.3 | 98 |
| 125 | Emerging concepts in the therapy of mitochondrial disease. Biochimica Et Biophysica Acta - Bioenergetics, 2015, 1847, 544-557. | 1.0 | 96 |
| 126 | GJA12 mutations in children with recessive hypomyelinating leukoencephalopathy. Neurology, 2006, 67, 273-279. | 1.1 | 95 |

| # | Article | IF | Citations |
|---|---|---|---|
| 127 | Neural stem cells traffic functional mitochondria via extracellular vesicles. PLoS Biology, 2021, 19, e3001166. | 5.6 | 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. | 2.5 | 91 |
| 130 | RNASEH1 Mutations Impair mtDNA Replication and Cause Adult-Onset Mitochondrial Encephalomyopathy. American Journal of Human Genetics, 2015, 97, 186-193. | 6.2 | 91 |
| 131 | Microscale oxygraphy reveals OXPHOS impairment in MRC mutant cells. Mitochondrion, 2012, 12, 328-335. | 3.4 | 90 |
| 132 | Complex IV-deficient Surf1â^'/â^' mice initiate mitochondrial stress responses. Biochemical Journal, 2014, 462, 359-371. | 3.7 | 89 |
| 133 | Disease-Causing SDHAF1 Mutations Impair Transfer of Fe-S Clusters to SDHB. Cell Metabolism, 2016, 23, 292-302. | 16.2 | 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. | 7.7 | 89 |
| 135 | An autosomal locus predisposing to multiple deletions of mtDNA on chromosome 3p. American Journal of Human Genetics, 1996, 58, 763-9. | 6.2 | 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. | 2.9 | 87 |
| 137 | Early-onset liver mtDNA depletion and late-onset proteinuric nephropathy in Mpv17 knockout mice. Human Molecular Genetics, 2009, 18, 12-26. | 2.9 | 87 |
| 138 | New genes and pathomechanisms in mitochondrial disorders unraveled by NGS technologies. Biochimica Et Biophysica Acta - Bioenergetics, 2016, 1857, 1326-1335. | 1.0 | 87 |
| 139 | VARS2andTARS2Mutations in Patients with Mitochondrial Encephalomyopathies. Human Mutation, 2014, 35, 983-989. | 2.5 | 86 |
| 140 | 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 |
| 141 | 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 |
| 142 | Rapamycin rescues mitochondrial myopathy via coordinated activation of autophagy and lysosomal biogenesis. EMBO Molecular Medicine, 2018, 10, . | 6.9 | 86 |
| 143 | A novel missense adenine nucleotide translocator-1 gene mutation in a Greek adPEO family. Neurology, 2001, 57, 2295-2298. | 1.1 | 85 |
| 144 | Depletion of mtDNA: Syndromes and genes. Mitochondrion, 2007, 7, 6-12. | 3.4 | 85 |

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|---|---|---|---|
| 145 | Respiratory chain and mitochondrial DNA in muscle and brain in Parkinson's disease patients. Neurology, 1993, 43, 2262-2262. | 1.1 | 85 |
| 146 | Benign reversible muscle cytochrome c oxidase deficiency. Neurology, 1987, 37, 64-64. | 1.1 | 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. | 2.9 | 83 |
| 148 | Mitochondrial Disorders. Medicine (United States), 1998, 77, 59-72. | 1.0 | 83 |
| 149 | Identification of novel mutations in five patients with mitochondrial encephalomyopathy. Biochimica Et Biophysica Acta - Bioenergetics, 2009, 1787, 491-501. | 1.0 | 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. | 5.3 | 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. | 6.2 | 82 |
| 152 | Decreased aminoacylation of mutant tRNAs in MELAS but not in MERRF patients. Human Molecular Genetics, 2000, 9, 467-475. | 2.9 | 82 |
| 153 | Nuclear genes in mitochondrial disorders. Current Opinion in Genetics and Development, 2003, 13, 262-270. | 3.3 | 82 |
| 154 | Myofibrillar-protein isoforms and sarcoplasmic-reticulum Ca2+-transport activity of single human muscle fibres. Biochemical Journal, 1984, 224, 215-225. | 3.7 | 81 |
| 155 | Dysregulated mitophagy and mitochondrial organization in optic atrophy due to OPA1 mutations. Neurology, 2017, 88, 131-142. | 1.1 | 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. | 2.8 | 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. | 2.9 | 80 |
| 158 | Chronic Exposure to Sulfide Causes Accelerated Degradation of Cytochrome c Oxidase in Ethylmalonic Encephalopathy. Antioxidants and Redox Signaling, 2011, 15, 353-362. | 5.4 | 80 |
| 159 | Mitochondrial disorders. Current Neurology and Neuroscience Reports, 2003, 3, 423-432. | 4.2 | 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. | 7.9 | 79 |
| 161 | Fumarase deficiency is an autosomal recessive encephalopathy affecting both the mitochondrial and the cytosolic enzymes. Neurology, 1990, 40, 495-495. | 1.1 | 79 |
| 162 | Neurological presentations of mitochondrial diseases. Journal of Inherited Metabolic Disease, 1996, 19, 504-520. | 3.6 | 78 |

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|---|---|---|---|
| 163 | ETHE1 mutations are specific to ethylmalonic encephalopathy. Journal of Medical Genetics, 2005, 43, 340-346. | 3.2 | 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. | 3.2 | 78 |
| 165 | Loss of apoptosis-inducing factor critically affects MIA40 function. Cell Death and Disease, 2015, 6, e1814-e1814. | 6.3 | 77 |
| 166 | Myopathy and Fatal Cardiopathy due to Cytochrome c Oxidase Deficiency. Archives of Neurology, 1986, 43, 1198-1202. | 4.5 | 76 |
| 167 | Genotypes from patients indicate no paternal mitochondrial DNA contribution. Annals of Neurology, 2003, 54, 521-524. | 5.3 | 76 |
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