Lee-Jun C Wong

List of Publications by Year
in descending order

202 papers	10,202 citations	25034 57 h-index	45317 90 g-index
210 all docs	210 docs citations	210 times ranked	11948 citing authors

#	Article	IF	Citations
1	Clinical Spectrum, Morbidity, and Mortality in 113 Pediatric Patients With Mitochondrial Disease. Pediatrics, 2004, 114, 925-931.	2.1	431
2	Biparental Inheritance of Mitochondrial DNA in Humans. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 13039-13044.	7.1	349
3	The in-depth evaluation of suspected mitochondrial disease. Molecular Genetics and Metabolism, 2008, 94, 16-37.	1.1	320
4	Somatic mosaicism, germline expansions, germline reversions and intergenerational reductions in myotonic dystrophy males: small pool PCR analyses. Human Molecular Genetics, 1995, 4, 1-8.	2.9	312
5	Primary immunodeficiency diseases: Genomic approaches delineate heterogeneous Mendelian disorders. Journal of Allergy and Clinical Immunology, 2017, 139, 232-245.	2.9	261
6	Molecular and clinical genetics of mitochondrial diseases due toPOLGmutations. Human Mutation, 2008, 29, E150-E172.	2.5	256
7	Fatty Acid Oxidation-Driven Src Links Mitochondrial Energy Reprogramming and Oncogenic Properties in Triple-Negative Breast Cancer. Cell Reports, 2016, 14, 2154-2165.	6.4	232
8	Comprehensive scanning of somatic mitochondrial DNA mutations in breast cancer. Cancer Research, 2002, 62, 972-6.	0.9	192
9	Non-invasive prenatal sequencing for multiple Mendelian monogenic disorders using circulating cell-free fetal DNA. Nature Medicine, 2019, 25, 439-447.	30.7	160
10	Detection and Quantification of Heteroplasmic Mutant Mitochondrial DNA by Real-Time Amplification Refractory Mutation System Quantitative PCR Analysis: A Single-Step Approach. Clinical Chemistry, 2004, 50, 996-1001.	3.2	151
11	Comprehensive One-Step Molecular Analyses of Mitochondrial Genome by Massively Parallel Sequencing. Clinical Chemistry, 2012, 58, 1322-1331.	3.2	144
12	Mitochondrial DNA polymerase Â mutations: an ever expanding molecular and clinical spectrum. Journal of Medical Genetics, 2011, 48, 669-681.	3.2	140
13	Mutations in FBXL4, Encoding a Mitochondrial Protein, Cause Early-Onset Mitochondrial Encephalomyopathy. American Journal of Human Genetics, 2013, 93, 482-495.	6.2	138
14	Simultaneous Detection and Quantification of Mitochondrial DNA Deletion(s), Depletion, and Over-Replication in Patients with Mitochondrial Disease. Journal of Molecular Diagnostics, 2005, 7, 613-622.	2.8	137
15	Mutations in FBXL4 Cause Mitochondrial Encephalopathy and a Disorder of Mitochondrial DNA Maintenance. American Journal of Human Genetics, 2013, 93, 471-481.	6.2	137
16	POLG DNA testing as an emerging standard of care before instituting valproic acid therapy for pediatric seizure disorders. Seizure: the Journal of the British Epilepsy Association, 2010, 19, 140-146.	2.0	136
17	Good laboratory practice for clinical next-generation sequencing informatics pipelines. Nature Biotechnology, 2015, 33, 689-693.	17.5	134
18	Quantitative Evaluation of the Mitochondrial DNA Depletion Syndrome. Clinical Chemistry, 2010, 56, 1119-1127.	3.2	119

#	Article	IF	Citations
19	Quantitative PCR Analysis of Mitochondrial DNA Content in Patients with Mitochondrial Disease. Annals of the New York Academy of Sciences, 2004, 1011, 304-309.	3.8	112
20	Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE)-like phenotype: an expanded clinical spectrum of POLG1 mutations. Journal of Neurology, 2012, 259, 862-868.	3.6	112
21	Mutations in theMPV17 gene are responsible for rapidly progressive liver failure in infancy. Hepatology, 2007, 46, 1218-1227.	7.3	111
22	Pathogenic mitochondrial DNA mutations in protein-coding genes. Muscle and Nerve, 2007, 36, 279-293.	2.2	109
23	Comprehensive next-generation sequence analyses of the entire mitochondrial genome reveal new insights into the molecular diagnosis of mitochondrial DNA disorders. Genetics in Medicine, 2013, 15, 388-394.	2.4	106
24	Mitochondrial Neurogastrointestinal Encephalopathy Due to Mutations in RRM2B. Archives of Neurology, 2009, 66, 1028-32.	4.5	103
25	Maternal systemic primary carnitine deficiency uncovered by newborn screening: Clinical, biochemical, and molecular aspects. Genetics in Medicine, 2010, 12, 19-24.	2.4	91
26	Realâ€Time Quantitative PCR Analysis of Mitochondrial DNA Content. Current Protocols in Human Genetics, 2011, 68, Unit 19.7..	3.5	90
27	Cyclic vomiting syndrome and mitochondrial DNA mutations. Lancet, The, 1997, 350, 1299-1300.	13.7	88
28	Variable clinical manifestation of homoplasmic G14459A mitochondrial DNA mutation. American Journal of Medical Genetics Part A, 2004, 124A, 377-382.	2.4	87
29	Comprehensive Scanning of the Entire Mitochondrial Genome for Mutations. Clinical Chemistry, 2002, 48, 1901-1912.	3.2	85
30	Rapid molecular diagnostics of severe primary immunodeficiency determined by using targeted next-generation sequencing. Journal of Allergy and Clinical Immunology, 2016, 138, 1142-1151.e2.	2.9	85
31	Direct detection of multiple point mutations in mitochondrial DNA. Clinical Chemistry, 1997, 43, 1857-1861.	3.2	84
32	Yield of mtDNA mutation analysis in 2,000 patients. , 1998, 77, 395-400.		84
33	Mitochondrial DNA analysis in clinical laboratory diagnostics. Clinica Chimica Acta, 2005, 354, 1-20.	1.1	83
34	Detection of Mitochondrial DNA Mutations by Temporal Temperature Gradient Gel Electrophoresis. Clinical Chemistry, 1999, 45, 1162-1167.	3.2	82
35	Citrin Deficiency: A Novel Cause of Failure to Thrive That Responds to a High-Protein, Low-Carbohydrate Diet. Pediatrics, 2007, 119, e773-e777.	2.1	81
36	Citrin deficiency, a perplexing global disorder. Molecular Genetics and Metabolism, 2009, 96, 44-49.	1.1	81

#	Article	IF	Citations
37	MPV17-associated hepatocerebral mitochondrial DNA depletion syndrome: New patients and novel mutations. Molecular Genetics and Metabolism, 2010, 99, 300-308.	1.1	80
38	Transition to Next Generation Analysis of the Whole Mitochondrial Genome: A Summary of Molecular Defects. Human Mutation, 2013, 34, 882-893.	2.5	79
39	Utility of Oligonucleotide Arrayâ€"Based Comparative Genomic Hybridization for Detection of Target Gene Deletions. Clinical Chemistry, 2008, 54, 1141-1148.	3.2	78
40	Mitochondrial dysfunction in human breast cancer cells and their transmitochondrial cybrids. Biochimica Et Biophysica Acta - Bioenergetics, 2010, 1797, 29-37.	1.0	77
41	Crosstalk from Non-Cancerous Mitochondria Can Inhibit Tumor Properties of Metastatic Cells by Suppressing Oncogenic Pathways. PLoS ONE, 2013, 8, e61747.	2.5	76
42	Diagnostic challenges of mitochondrial DNA disorders. Mitochondrion, 2007, 7, 45-52.	3.4	75
43	Diagnosis of mitochondrial myopathies. Molecular Genetics and Metabolism, 2013, 110, 35-41.	1.1	74
44	Sensory ataxic neuropathy with ophthalmoparesis caused by POLG mutations. Neuromuscular Disorders, 2008, 18, 626-632.	0.6	71
45	Early-onset severe neuromuscular phenotype associated with compound heterozygosity for OPA1 mutations. Molecular Genetics and Metabolism, 2011, 103, 383-387.	1.1	71
46	Next generation molecular diagnosis of mitochondrial disorders. Mitochondrion, 2013, 13, 379-387.	3.4	71
47	The next generation of population-based spinal muscular atrophy carrier screening: comprehensive pan-ethnic SMN1 copy-number and sequence variant analysis by massively parallel sequencing. Genetics in Medicine, 2017, 19, 936-944.	2.4	70
48	Detection and Quantification of Mosaic Mutations in Disease Genes by Next-Generation Sequencing. Journal of Molecular Diagnostics, 2016, 18, 446-453.	2.8	69
49	Human mitochondrial transfer RNAs: Role of pathogenic mutation in disease. Muscle and Nerve, 2008, 37, 150-171.	2.2	68
50	Improved molecular diagnosis by the detection of exonic deletions with target gene capture and deep sequencing. Genetics in Medicine, 2015, 17, 99-107.	2.4	68
51	Abnormal neurological features predict poor survival and should preclude liver transplantation in patients with deoxyguanosine kinase deficiency. Liver Transplantation, 2008, 14, 1480-1485.	2.4	67
52	Dependable and Efficient Clinical Utility of Target Capture-Based Deep Sequencing in Molecular Diagnosis of Retinitis Pigmentosa. , 2014, 55, 6213.		67
53	Detection of mitochondrial DNA mutations in the tumor and cerebrospinal fluid of medulloblastoma patients. Cancer Research, 2003, 63, 3866-71.	0.9	67
54	Current molecular diagnostic algorithm for mitochondrial disorders. Molecular Genetics and Metabolism, 2010, 100, 111-117.	1.1	66

#	Article	IF	Citations
55	Clinical application of massively parallel sequencing in the molecular diagnosis of glycogen storage diseases of genetically heterogeneous origin. Genetics in Medicine, 2013, 15, 106-114.	2.4	65
56	Recurrent ACADVL molecular findings in individuals with a positive newborn screen for very long chain acyl-coA dehydrogenase (VLCAD) deficiency in the United States. Molecular Genetics and Metabolism, 2015, 116, 139-145.	1.1	65
57	Mitochondrial myopathy, lactic acidosis, and sideroblastic anemia (MLASA) plus associated with a novel de novo mutation (m.8969G>A) in the mitochondrial encoded ATP6 gene. Molecular Genetics and Metabolism, 2014, 113, 207-212.	1.1	63
58	Hepatocellular carcinoma associated with tightâ€junction protein 2 deficiency. Hepatology, 2015, 62, 1914-1916.	7.3	63
59	Molecular genetics of mitochondrial disorders. Developmental Disabilities Research Reviews, 2010, 16, 154-162.	2.9	62
60	Molecular spectrum of SLC22A5 (OCTN2) gene mutations detected in 143 subjects evaluated for systemic carnitine deficiency. Human Mutation, 2010, 31, E1632-E1651.	2.5	61
61	Biochemical analysis of human POLG2 variants associated with mitochondrial disease. Human Molecular Genetics, 2011, 20, 3052-3066.	2.9	57
62	Arginine:glycine amidinotransferase (AGAT) deficiency: Clinical features and long term outcomes in 16 patients diagnosed worldwide. Molecular Genetics and Metabolism, 2015, 116, 252-259.	1.1	55
63	Finding twinkle in the eyes of a 71â€yearâ€old lady: A case report and review of the genotypic and phenotypic spectrum of TWINKLEâ€related dominant disease. American Journal of Medical Genetics, Part A, 2009, 149A, 861-867.	1.2	53
64	Mitochondrial DNA Content Varies with Pathological Characteristics of Breast Cancer. Journal of Oncology, 2011, 2011, 1-10.	1.3	52
65	Mitochondrial Syndromes with Leukoencephalopathies. Seminars in Neurology, 2012, 32, 055-061.	1.4	52
66	Application of dual-genome oligonucleotidearray-based comparative genomic hybridization to the molecular diagnosis of mitochondrial DNA deletion and depletion syndromes. Genetics in Medicine, 2009, 11, 518-526.	2.4	51
67	A Founder Mutation in VPS11 Causes an Autosomal Recessive Leukoencephalopathy Linked to Autophagic Defects. PLoS Genetics, 2016, 12, e1005848.	3.5	50
68	Alternative, Noninvasive Tissues for Quantitative Screening of Mutant Mitochondrial DNA. Clinical Chemistry, 1997, 43, 1241-1243.	3.2	49
69	Mitochondrial DNA variant interactions modify breast cancer risk. Journal of Human Genetics, 2008, 53, 924-928.	2.3	49
70	Sequence Homology at the Breakpoint and Clinical Phenotype of Mitochondrial DNA Deletion Syndromes. PLoS ONE, 2010, 5, e15687.	2.5	49
71	Improved detection ofCFTR mutations in Southern California Hispanic CF patients. Human Mutation, 2001, 18, 296-307.	2.5	48
72	High-frequency detection of deletions and variable rearrangements at the ornithine transcarbamylase (OTC) locus by oligonucleotide array CGH. Molecular Genetics and Metabolism, 2009, 96, 97-105.	1.1	48

#	Article	IF	Citations
73	Expanding genotype/phenotype of neuromuscular diseases by comprehensive target capture/NGS. Neurology: Genetics, 2015, 1, e14.	1.9	48
74	The mitochondrial 13513G>A mutation is associated with Leigh disease phenotypes independent of complex I deficiency in muscle. Molecular Genetics and Metabolism, 2008, 94, 485-490.	1.1	47
75	MPV17-related mitochondrial DNA maintenance defect: New cases and review of clinical, biochemical, and molecular aspects. Human Mutation, 2018, 39, 461-470.	2.5	45
76	Somatic Mitochondrial DNA Mutations in Neurofibromatosis Type 1-Associated Tumors. Molecular Cancer Research, 2004, 2, 433-441.	3.4	45
77	Macular Pattern Retinal Dystrophy, Adult-onset Diabetes, and Deafness: A Family Study of A3243G Mitochondrial Heteroplasmy. American Journal of Ophthalmology, 1997, 124, 217-221.	3.3	44
78	Application of Next Generation Sequencing to Molecular Diagnosis of Inherited Diseases. Topics in Current Chemistry, 2012, 336, 19-45.	4.0	43
79	MIPEP recessive variants cause a syndrome of left ventricular non-compaction, hypotonia, and infantile death. Genome Medicine, 2016, 8, 106.	8.2	43
80	Clinical Heterogeneity in Mitochondrial DNA Deletion Disorders: A Diagnostic Challenge of Pearson Syndrome. American Journal of Medical Genetics Part A, 2000, 95, 266-268.	2.4	42
81	A Comprehensive Strategy for Accurate Mutation Detection of the Highly Homologous PMS2. Journal of Molecular Diagnostics, 2015, 17, 545-553.	2.8	42
82	Mitochondria of highly metastatic breast cancer cell line MDA-MB-231 exhibits increased autophagic properties. Biochimica Et Biophysica Acta - Bioenergetics, 2011, 1807, 1125-1132.	1.0	41
83	ADIPOR1Is Mutated in Syndromic Retinitis Pigmentosa. Human Mutation, 2016, 37, 246-249.	2.5	41
84	Molecular and clinical spectra of FBXL4 deficiency. Human Mutation, 2017, 38, 1649-1659.	2.5	41
85	SURF1-associated leigh syndrome: A case series and novel mutations. Human Mutation, 2012, 33, 1192-1200.	2.5	40
86	Molecular and clinical characterization of the myopathic form of mitochondrial DNA depletion syndrome caused by mutations in the thymidine kinase (TK2) gene. Molecular Genetics and Metabolism, 2013, 110, 153-161.	1.1	40
87	An integrated approach for classifying mitochondrial DNA variants: one clinical diagnostic laboratoryâ€™s experience. Genetics in Medicine, 2012, 14, 620-626.	2.4	39
88	Molecular analysis for mitochondrial DNA disorders. Mitochondrion, 2004, 4, 403-415.	3.4	38
89	Targeted array CGH as a valuable molecular diagnostic approach: Experience in the diagnosis of mitochondrial and metabolic disorders. Molecular Genetics and Metabolism, 2012, 106, 221-230.	1.1	38
90	Diagnostic approaches to apparent homozygosity. Genetics in Medicine, 2012, 14, 877-882.	2.4	38

#	Article	IF	Citations
91	Mitochondrial disorder with OPA1 mutation lacking optic atrophy. Mitochondrion, 2009, 9, 279-281.	3.4	37
92	Î²2-adrenergic receptors mediate cardioprotection through crosstalk with mitochondrial cell death pathways. Journal of Molecular and Cellular Cardiology, 2011, 51, 781-789.	1.9	37
93	Severe lactic acidosis caused by a novel frame-shift mutation in mitochondrial-encoded cytochrome c oxidase subunit II. American Journal of Medical Genetics Part A, 2001, 102, 95-99.	2.4	35
94	Interpretation of mitochondrial tRNA variants. Genetics in Medicine, 2020, 22, 917-926.	2.4	35
95	Molecularâ€“clinical correlations in a family with variable tissue mitochondrial DNA T8993G mutant load. Molecular Genetics and Metabolism, 2006, 88, 364-371.	1.1	34
96	Functional effects of cancer mitochondria on energy metabolism and tumorigenesis: utility of transmitochondrial cybrids. Annals of the New York Academy of Sciences, 2010, 1201, 137-146.	3.8	34
97	Small heterodimer partner deletion prevents hepatic steatosis and when combined with farnesoid X receptor loss protects against type 2 diabetes in mice. Hepatology, 2017, 66, 1854-1865.	7.3	34
98	Loss of Oxidation Resistance 1, OXR1, Is Associated with an Autosomal-Recessive Neurological Disease with Cerebellar Atrophy and Lysosomal Dysfunction. American Journal of Human Genetics, 2019, 105, 1237-1253.	6.2	34
99	Deoxyguanosine kinase deficiency presenting as neonatal hemochromatosis. Molecular Genetics and Metabolism, 2011, 103, 262-267.	1.1	33
100	Detection of mitochondrial DNA mutations using temporal temperature gradient gel electrophoresis. Electrophoresis, 2004, 25, 2602-2610.	2.4	32
101	Clinical and molecular spectrum of thymidine kinase 2-related mtDNA maintenance defect. Molecular Genetics and Metabolism, 2018, 124, 124-130.	1.1	32
102	Somatic Mitochondrial DNA Mutations in Oral Cancer of Betel Quid Chewers. Annals of the New York Academy of Sciences, 2004, 1011, 310-316.	3.8	31
103	POLG-related disorders. Neurology, 2011, 77, 1847-1852.	1.1	31
104	A cystic fibrosis patient with two novel mutations in mitochondrial DNA: Mild disease led to delayed diagnosis of both disorders. American Journal of Medical Genetics Part A, 2002, 113, 59-64.	2.4	30
105	Elevations of C14:1 and C14:2 Plasma Acylcarnitines in Fasted Children: AÂDiagnostic Dilemma. Journal of Pediatrics, 2016, 169, 208-213.e2.	1.8	30
106	Comprehensive scanning of the entire mitochondrial genome for mutations. Clinical Chemistry, 2002, 48, 1901-12.	3.2	30
107	Novel POLG Splice Site Mutation and Optic Atrophy. Archives of Neurology, 2011, 68, 806-11.	4.5	29
108	Recognition of mitochondrial DNA deletion syndrome with non-neuromuscular multisystemic manifestation. Genetics in Medicine, 2001, 3, 399-404.	2.4	28

#	Article	IF	Citations
109	Two mtDNA mutations 14487T>C (M63V, ND6) and 12297T>C (tRNA Leu) in a Leigh syndrome family. Molecular Genetics and Metabolism, 2009, 96, 59-65.	1.1	28
110	FARS2 deficiency; new cases, review of clinical, biochemical, and molecular spectra, and variants interpretation based on structural, functional, and evolutionary significance. Molecular Genetics and Metabolism, 2018, 125, 281-291.	1.1	28
111	Expanded molecular features of carnitine acyl-carnitine translocase (CACT) deficiency by comprehensive molecular analysis. Molecular Genetics and Metabolism, 2011, 103, 349-357.	1.1	27
112	Comprehensive Mitochondrial Genome Analysis by Massively Parallel Sequencing. Methods in Molecular Biology, 2016, 1351, 3-17.	0.9	26
113	Analysis of Mitochondrial DNA Point Mutation Heteroplasmy by ARMS Quantitative PCR. Current Protocols in Human Genetics, 2011, 68, Unit 19.6..	3.5	25
114	Two patients with hepatic mtDNA depletion syndromes and marked elevations of S-adenosylmethionine and methionine. Molecular Genetics and Metabolism, 2012, 105, 228-236.	1.1	25
115	Atypical presentation of Leigh syndrome associated with a Leber hereditary optic neuropathy primary mitochondrial DNA mutation. Molecular Genetics and Metabolism, 2011, 103, 153-160.	1.1	24
116	Challenges of Bringing Next Generation Sequencing Technologies to Clinical Molecular Diagnostic Laboratories. Neurotherapeutics, 2013, 10, 262-272.	4.4	24
117	Congenital heart defects and left ventricular non-compaction in males with loss-of-function variants in<i>NONO</i>. Journal of Medical Genetics, 2017, 54, 47-53.	3.2	24
118	Succinyl-CoA synthetase (SUCLA2) deficiency in two siblings with impaired activity of other mitochondrial oxidative enzymes in skeletal muscle without mitochondrial DNA depletion. Molecular Genetics and Metabolism, 2017, 120, 213-222.	1.1	24
119	Added value of next generation gene panel analysis for patients with elevated methylmalonic acid and no clinical diagnosis following functional studies of vitamin B12 metabolism. Molecular Genetics and Metabolism, 2016, 117, 363-368.	1.1	23
120	Capture-based high-coverage NGS: a powerful tool to uncover a wide spectrum of mutation types. Genetics in Medicine, 2016, 18, 513-521.	2.4	23
121	Preparation and Validation of PCR-generated Positive Controls for Diagnostic Dot Blotting. Clinical Chemistry, 1998, 44, 1578-1579.	3.2	22
122	Novel mitochondrial DNA mutations associated with myopathy, cardiomyopathy, renal failure, and deafness. American Journal of Medical Genetics, Part A, 2006, 140A, 2216-2222.	1.2	22
123	Multiplex Ligation-Dependent Probe Amplification Identification of Whole Exon and Single Nucleotide Deletions in the CFTR Gene of Hispanic Individuals with Cystic Fibrosis. Journal of Molecular Diagnostics, 2008, 10, 368-375.	2.8	22
124	Novel insights into the functional metabolic impact of an apparent de novo m.8993T>G variant in the MT-ATP6 gene associated with maternally inherited form of Leigh Syndrome. Molecular Genetics and Metabolism, 2018, 124, 71-81.	1.1	22
125	Application of oligonucleotide array CGH to the simultaneous detection of a deletion in the nuclear TK2 gene and mtDNA depletion. Molecular Genetics and Metabolism, 2010, 99, 53-57.	1.1	21
126	Detection of uniparental isodisomy in autosomal recessive mitochondrial DNA depletion syndrome by high-density SNP array analysis. Journal of Human Genetics, 2011, 56, 834-839.	2.3	21

#	Article	IF	Citations
127	Improved Diagnosis of Inherited Retinal Dystrophies by High-Fidelity PCR of ORF15 followed by Next-Generation Sequencing. Journal of Molecular Diagnostics, 2016, 18, 817-824.	2.8	21
128	The phenotypic variability of HK1-associated retinal dystrophy. Scientific Reports, 2017, 7, 7051.	3.3	21
129	Comprehensive target capture/next-generation sequencing as a second-tier diagnostic approach for congenital muscular dystrophy in Taiwan. PLoS ONE, 2017, 12, e0170517.	2.5	21
130	Intergenerational transmission of pathogenic heteroplasmic mitochondrial DNA. Genetics in Medicine, 2002, 4, 78-83.	2.4	20
131	Leigh syndrome caused by a novel m.4296G>A mutation in mitochondrial tRNA isoleucine. Mitochondrion, 2012, 12, 258-261.	3.4	20
132	Comprehensive Molecular Diagnosis of Mitochondrial Disorders: Qualitative and Quantitative Approach. Annals of the New York Academy of Sciences, 2004, 1011, 246-258.	3.8	19
133	Exercise intolerance due to cytochrome b mutation. Muscle and Nerve, 2010, 42, 136-140.	2.2	19
134	Reduced Mitochondrial DNA Content and Heterozygous Nuclear Gene Mutations in Patients With Acute Liver Failure. Journal of Pediatric Gastroenterology and Nutrition, 2013, 57, 438-443.	1.8	18
135	A SUCLG1 mutation in a patient with mitochondrial DNA depletion and congenital anomalies. Molecular Genetics and Metabolism Reports, 2014, 1, 451-454.	1.1	18
136	Adult-onset respiratory insufficiency, scoliosis, and distal joint hyperlaxity in patients with multiminicore disease due to novel Megf10 mutations. Muscle and Nerve, 2016, 53, 984-988.	2.2	18
137	Novel SEA and LG2 Agrin mutations causing congenital Myasthenic syndrome. Orphanet Journal of Rare Diseases, 2017, 12, 182.	2.7	18
138	Alpers syndrome with prominent white matter changes. Brain and Development, 2008, 30, 295-300.	1.1	17
139	Progressive myofiber loss with extensive fibro-fatty replacement in a child with mitochondrial DNA depletion syndrome and novel thymidine kinase 2 gene mutations. Neuromuscular Disorders, 2009, 19, 784-787.	0.6	17
140	Fatal infantile lactic acidosis and a novel homozygous mutation in the SUCLG1 gene: A mitochondrial DNA depletion disorder. Molecular Genetics and Metabolism, 2011, 102, 149-152.	1.1	17
141	The necessity of complete mutational analysis of an infertile couple before in vitro fertilization. Fertility and Sterility, 2004, 82, 947-949.	1.0	16
142	Next generation deep sequencing corrects diagnostic pitfalls of traditional molecular approach in a patient with prenatal onset of Pompe disease. American Journal of Medical Genetics, Part A, 2017, 173, 2500-2504.	1.2	16
143	The nuclear background influences the penetrance of the near-homoplasmic m.1630 Aâ€¯>â€¯G MELAS variant in a symptomatic proband and asymptomatic mother. Molecular Genetics and Metabolism, 2019, 126, 429-438.	1.1	16
144	Detection ofCFTR mutations using temporal temperature gradient gel electrophoresis. Electrophoresis, 2004, 25, 2593-2601.	2.4	15

#	Article	IF	Citations
145	POLG mutation in a patient with cataracts, early-onset distal muscle weakness and atrophy, ovarian dysgenesis and 3-methylglutaconic aciduria. Gene, 2012, 499, 209-212.	2.2	15
146	Reply to Lutz-Bonengel et al.: Biparental mtDNA transmission is unlikely to be the result of nuclear mitochondrial DNA segments. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 1823-1824.	7.1	15
147	Novel SNP at the common primer site of exon IIIa ofFGFR2 gene causes error in molecular diagnosis of craniosynostosis syndrome. American Journal of Medical Genetics Part A, 2001, 102, 282-285.	2.4	14
148	Detection of a novel intragenic rearrangement in the creatine transporter gene by next generation sequencing. Molecular Genetics and Metabolism, 2013, 110, 465-471.	1.1	14
149	Axial mitochondrial myopathy in a patient with rapidly progressive adult-onset scoliosis. Acta Neuropathologica Communications, 2014, 2, 137.	5.2	14
150	Mitochondrial inheritance and cancer. Translational Research, 2018, 202, 24-34.	5.0	14
151	PRENATAL DIAGNOSIS OF GLYCOGEN STORAGE DISEASE TYPE 1a BY DIRECT MUTATION DETECTION. , 1996, 16, 105-108.		13
152	A novel mutation detected by temporal temperature gradient gel electrophoresis led to the confirmative prenatal diagnosis of a Hispanic CF family. Prenatal Diagnosis, 2000, 20, 807-810.	2.3	13
153	AIB1 gene amplification and the instability of polyQ encoding sequence in breast cancer cell lines. BMC Cancer, 2006, 6, 111.	2.6	13
154	A novel c.592-4_c.592-3delTT mutation in DGUOK gene causes exon skipping. Mitochondrion, 2010, 10, 188-191.	3.4	13
155	Somatic mosaicism for PDHA1 mutation in a male with pyruvate dehydrogenase complex deficiency. Molecular Genetics and Metabolism, 2010, 100, 296-299.	1.1	13
156	Kinetic and structural changes in <scp>H</scp>smt<scp>P</scp>he<scp>RS</scp>, induced by pathogenic mutations in human <scp>FARS</scp>2. Protein Science, 2017, 26, 1505-1516.	7.6	13
157	Application of oligonucleotide array CGH in the detection of a large intragenic deletion in POLG associated with Alpers Syndrome. Mitochondrion, 2011, 11, 104-107.	3.4	12
158	Clinical and laboratory interpretation of mitochondrial mRNA variants. Human Mutation, 2020, 41, 1783-1796.	2.5	12
159	Real-Time Quantitative Polymerase Chain Reaction Analysis of Mitochondrial DNA Point Mutation. , 2006, 335, 187-200.		11
160	De Novo Mutations in POLG Presenting with Acute Liver Failure or Encephalopathy. Journal of Pediatric Gastroenterology and Nutrition, 2009, 49, 126-129.	1.8	11
161	Molecular characterization of CPS1 deletions by array CGH. Molecular Genetics and Metabolism, 2011, 102, 103-106.	1.1	11
162	Mitochondrial myopathy due to novel missense mutation in the cytochrome c oxidase 1 gene. Journal of the Neurological Sciences, 2012, 319, 158-163.	0.6	11

#	Article	IF	Citations
163	Molecular Analysis of Mitochondrial DNA Point Mutations by Polymerase Chain Reaction. , 2006, 336, 135-144.		10
164	Disclosing the functional changes of two genetic alterations in a patient with Chronic Progressive External Ophthalmoplegia: Report of the novel mtDNA m.7486G>A variant. Neuromuscular Disorders, 2018, 28, 350-360.	0.6	10
165	Clinical Utility of Rapid Exome Sequencing Combined With Mitochondrial DNA Sequencing in Critically Ill Pediatric Patients With Suspected Genetic Disorders. Frontiers in Genetics, 2021, 12, 725259.	2.3	10
166	Exercise intolerance associated with a novel 8300t>C mutation in mitochondrial transfer RNAlys. Muscle and Nerve, 2006, 34, 437-443.	2.2	9
167	Utilization of targeted array comparative genomic hybridization, MitoMetÂ®, in prenatal diagnosis of metabolic disorders. Molecular Genetics and Metabolism, 2011, 103, 148-152.	1.1	9
168	Next generation sequencing of patients with mut methylmalonic aciduria: Validation of somatic cell studies and identification of 16 novel mutations. Molecular Genetics and Metabolism, 2016, 118, 264-271.	1.1	9
169	Retinal Diseases Caused by Mutations in Genes Not Specifically Associated with the Clinical Diagnosis. PLoS ONE, 2016, 11, e0165405.	2.5	9
170	A mitochondrial DNA mutation in a patient with an extensive family history of Duchenne muscular dystrophy. Muscle and Nerve, 2004, 30, 118-122.	2.2	8
171	Expanding the clinical phenotype of the mitochondrial m.13513G>A mutation with the first report of a fatal neonatal presentation. Developmental Medicine and Child Neurology, 2011, 53, 565-568.	2.1	8
172	Myopathy during treatment with the antianginal drug ranolazine. Journal of the Neurological Sciences, 2014, 347, 380-382.	0.6	8
173	The m.11778 Aâ€˜>â€˜G variant associated with the coexistence of Leber's hereditary optic neuropathy and multiple sclerosis-like illness dysregulates the metabolic interplay between mitochondrial oxidative phosphorylation and glycolysis. Mitochondrion, 2019, 46, 187-194.	3.4	7
174	Dystrophinopathy mimicking metabolic myopathies. Neuromuscular Disorders, 2015, 25, 653-657.	0.6	6
175	A novel Dral polymorphism in the 3â€˜ untranslated region of human glucoseâ€6â€phosphatase gene: useful for carrier detection and prenatal diagnosis of glycogen storage disease type 1a. Clinical Genetics, 1998, 53, 502-503.	2.0	5
176	Abnormalities in Glycogen Metabolism in a Patient with Alpersâ€™ Syndrome Presenting with Hypoglycemia. JIMD Reports, 2013, 14, 29-35.	1.5	5
177	Molecular genetic and mitochondrial metabolic analyses confirm the suspected mitochondrial etiology in a pediatric patient with an atypical form of alternating hemiplegia of childhood. Molecular Genetics and Metabolism Reports, 2020, 24, 100609.	1.1	5
178	Linking newborn severe combined immunodeficiency screening with targeted exome sequencing: A case report. Journal of Allergy and Clinical Immunology: in Practice, 2017, 5, 1442-1444.	3.8	4
179	The mitochondrial DNA variant m.9032TÂ>ÂC in MT-ATP6 encoding p.(Leu169Pro) causes a complex mitochondrial neurological syndrome. Mitochondrion, 2020, 55, 8-13.	3.4	4
180	Mitochondrial tRNA-serine (AGY) m.C12264T mutation causes severe multisystem disease with cataracts. Discovery Medicine, 2012, 13, 143-50.	0.5	4

#	Article	IF	Citations
181	Clinical Molecular Diagnostic Techniques: A Brief Review. , 2013, , 19-36.		3
182	Early onset and severe clinical course associated with the m.5540G>A mutation in MT - TW. Molecular Genetics and Metabolism Reports, 2014, 1, 61-65.	1.1	3
183	A novel acceptor stem variant in mitochondrial tRNATyr impairs mitochondrial translation and is associated with a severe phenotype. Molecular Genetics and Metabolism, 2020, 131, 398-404.	1.1	3
184	Genetic and Mitochondrial Metabolic Analyses of an Atypical Form of Leigh Syndrome. Frontiers in Cell and Developmental Biology, 2021, 9, 767407.	3.7	3
185	Two novel frame shift mutations ofCFTR causing null alleles in a patient with a severe course of CF. American Journal of Medical Genetics Part A, 2001, 102, 389-390.	2.4	2
186	Enhanced Detection of Deleterious Mutations by TTGE Analysis of Mother and Child's DNA Side by Side. Annals of the New York Academy of Sciences, 2004, 1011, 299-303.	3.8	2
187	Detection of Copy Number Variations (CNVs) Based on the Coverage Depth from the Next Generation Sequencing Data. , 2017, , 13-22.		2
188	Biochemical and Molecular Methods for the Study of Mitochondrial Disorders. , 2013, , 27-45.		1
189	Next-Generation Sequencing Analyses of the Whole Mitochondrial Genome. , 2013, , 203-219.		1
190	Novel OPA1 mutation featuring spastic paraparesis and intestinal dysmotility. Molecular Genetics and Metabolism Reports, 2014, 1, 443-445.	1.1	1
191	Overview of the Clinical Utility of Next Generation Sequencing in Molecular Diagnoses of Human Genetic Disorders. , 2017, , 1-11.		1
192	Extra-muscular manifestations of TK2 deficiency. Molecular Genetics and Metabolism Reports, 2018, 16, 30.	1.1	1
193	Yield of mtDNA mutation analysis in 2,000 patients. American Journal of Medical Genetics Part A, 1998, 77, 395-400.	2.4	1
194	Diagnostic approaches to apparent homozygosity. , 0, .		1
195	In reply to: Proving pathogenicity?when evolution is not enough. American Journal of Medical Genetics Part A, 2004, 131A, 109-110.	2.4	0
196	First prenatal exclusion of cystic fibrosis in East Asia. Pediatrics International, 2007, 49, 686-687.	0.5	0
197	Comprehensive Analyses of the Mitochondrial Genome. , 2017, , 287-304.		0
198	Next Generation Sequencing (NGS) Based Panel Analysis of Metabolic Pathways. , 2017, , 23-49.		0

#	Article	IF	Citations
199	Response to Bai et al.. Genetics in Medicine, 2020, 22, 1420-1421.	2.4	0
200	Mitochondrial immunofluorescence assay as an adjunctive tool in the diagnosis of mitochondrial myopathy. FASEB Journal, 2008, 22, 708.20.	0.5	0
201	Hereditary Paraganglioma and Pheochromocytoma. , 2016, , 393-399.		0
202	Molecular genetics of common mitochondrial DNA disorders. Beijing Da Xue Xue Bao, 2005, 37, 26-31.	0.2	0