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 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
2	Genetic and Mitochondrial Metabolic Analyses of an Atypical Form of Leigh Syndrome. Frontiers in Cell and Developmental Biology, 2021, 9, 767407.	3.7	3
3	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
4	Response to Bai et al.. Genetics in Medicine, 2020, 22, 1420-1421.	2.4	0
5	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
6	Clinical and laboratory interpretation of mitochondrial mRNA variants. Human Mutation, 2020, 41, 1783-1796.	2.5	12
7	Interpretation of mitochondrial tRNA variants. Genetics in Medicine, 2020, 22, 917-926.	2.4	35
8	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
9	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
10	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
11	Non-invasive prenatal sequencing for multiple Mendelian monogenic disorders using circulating cell-free fetal DNA. Nature Medicine, 2019, 25, 439-447.	30.7	160
12	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
13	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
14	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
15	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
16	Clinical and molecular spectrum of thymidine kinase 2-related mtDNA maintenance defect. Molecular Genetics and Metabolism, 2018, 124, 124-130.	1.1	32
17	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
18	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

#	Article	IF	Citations
19	Extra-muscular manifestations of TK2 deficiency. Molecular Genetics and Metabolism Reports, 2018, 16, 30.	1.1	1
20	Mitochondrial inheritance and cancer. Translational Research, 2018, 202, 24-34.	5.0	14
21	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
22	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
23	Congenital heart defects and left ventricular non-compaction in males with loss-of-function variants inNONO. Journal of Medical Genetics, 2017, 54, 47-53.	3.2	24
24	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
25	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
26	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
27	Overview of the Clinical Utility of Next Generation Sequencing in Molecular Diagnoses of Human Genetic Disorders. , 2017, , 1-11.		1
28	Comprehensive Analyses of the Mitochondrial Genome. , 2017, , 287-304.		0
29	Next Generation Sequencing (NGS) Based Panel Analysis of Metabolic Pathways. , 2017, , 23-49.		0
30	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
31	Molecular and clinical spectra of FBXL4 deficiency. Human Mutation, 2017, 38, 1649-1659.	2.5	41
32	The phenotypic variability of HK1-associated retinal dystrophy. Scientific Reports, 2017, 7, 7051.	3.3	21
33	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
34	Primary immunodeficiency diseases: Genomic approaches delineate heterogeneous Mendelian disorders. Journal of Allergy and Clinical Immunology, 2017, 139, 232-245.	2.9	261
35	Novel SEA and LG2 Agrin mutations causing congenital Myasthenic syndrome. Orphanet Journal of Rare Diseases, 2017, 12, 182.	2.7	18
36	Detection of Copy Number Variations (CNVs) Based on the Coverage Depth from the Next Generation Sequencing Data. , 2017, , 13-22.		2

#	Article	IF	Citations
37	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
38	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
39	MIPEP recessive variants cause a syndrome of left ventricular non-compaction, hypotonia, and infantile death. Genome Medicine, 2016, 8, 106.	8.2	43
40	ADIPOR1Is Mutated in Syndromic Retinitis Pigmentosa. Human Mutation, 2016, 37, 246-249.	2.5	41
41	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
42	Detection and Quantification of Mosaic Mutations in Disease Genes by Next-Generation Sequencing. Journal of Molecular Diagnostics, 2016, 18, 446-453.	2.8	69
43	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
44	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
45	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
46	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
47	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
48	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
49	Comprehensive Mitochondrial Genome Analysis by Massively Parallel Sequencing. Methods in Molecular Biology, 2016, 1351, 3-17.	0.9	26
50	A Founder Mutation in VPS11 Causes an Autosomal Recessive Leukoencephalopathy Linked to Autophagic Defects. PLoS Genetics, 2016, 12, e1005848.	3.5	50
51	Retinal Diseases Caused by Mutations in Genes Not Specifically Associated with the Clinical Diagnosis. PLoS ONE, 2016, 11, e0165405.	2.5	9
52	Hereditary Paraganglioma and Pheochromocytoma. , 2016, , 393-399.		0
53	Expanding genotype/phenotype of neuromuscular diseases by comprehensive target capture/NGS. Neurology: Genetics, 2015, 1, e14.	1.9	48
54	Hepatocellular carcinoma associated with tightâ€junction protein 2 deficiency. Hepatology, 2015, 62, 1914-1916.	7.3	63

#	Article	IF	Citations
55	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
56	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
57	Good laboratory practice for clinical next-generation sequencing informatics pipelines. Nature Biotechnology, 2015, 33, 689-693.	17.5	134
58	Dystrophinopathy mimicking metabolic myopathies. Neuromuscular Disorders, 2015, 25, 653-657.	0.6	6
59	A Comprehensive Strategy for Accurate Mutation Detection of the Highly Homologous PMS2. Journal of Molecular Diagnostics, 2015, 17, 545-553.	2.8	42
60	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
61	Dependable and Efficient Clinical Utility of Target Capture-Based Deep Sequencing in Molecular Diagnosis of Retinitis Pigmentosa. , 2014, 55, 6213.		67
62	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
63	Myopathy during treatment with the antianginal drug ranolazine. Journal of the Neurological Sciences, 2014, 347, 380-382.	0.6	8
64	Axial mitochondrial myopathy in a patient with rapidly progressive adult-onset scoliosis. Acta Neuropathologica Communications, 2014, 2, 137.	5.2	14
65	Novel OPA1 mutation featuring spastic paraparesis and intestinal dysmotility. Molecular Genetics and Metabolism Reports, 2014, 1, 443-445.	1.1	1
66	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
67	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
68	Challenges of Bringing Next Generation Sequencing Technologies to Clinical Molecular Diagnostic Laboratories. Neurotherapeutics, 2013, 10, 262-272.	4.4	24
69	Mutations in FBXL4, Encoding a Mitochondrial Protein, Cause Early-Onset Mitochondrial Encephalomyopathy. American Journal of Human Genetics, 2013, 93, 482-495.	6.2	138
70	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
71	Diagnosis of mitochondrial myopathies. Molecular Genetics and Metabolism, 2013, 110, 35-41.	1.1	74
72	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

#	Article	IF	Citations
73	Biochemical and Molecular Methods for the Study of Mitochondrial Disorders. , 2013, , 27-45.		1
74	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
75	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
76	Next generation molecular diagnosis of mitochondrial disorders. Mitochondrion, 2013, 13, 379-387.	3.4	71
77	Transition to Next Generation Analysis of the Whole Mitochondrial Genome: A Summary of Molecular Defects. Human Mutation, 2013, 34, 882-893.	2.5	79
78	Clinical Molecular Diagnostic Techniques: A Brief Review. , 2013, , 19-36.		3
79	Next-Generation Sequencing Analyses of the Whole Mitochondrial Genome. , 2013, , 203-219.		1
80	Abnormalities in Glycogen Metabolism in a Patient with Alpersâ€™ Syndrome Presenting with Hypoglycemia. JIMD Reports, 2013, 14, 29-35.	1.5	5
81	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
82	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
83	Crosstalk from Non-Cancerous Mitochondria Can Inhibit Tumor Properties of Metastatic Cells by Suppressing Oncogenic Pathways. PLoS ONE, 2013, 8, e61747.	2.5	76
84	Mitochondrial Syndromes with Leukoencephalopathies. Seminars in Neurology, 2012, 32, 055-061.	1.4	52
85	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
86	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
87	Leigh syndrome caused by a novel m.4296G>A mutation in mitochondrial tRNA isoleucine. Mitochondrion, 2012, 12, 258-261.	3.4	20
88	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
89	An integrated approach for classifying mitochondrial DNA variants: one clinical diagnostic laboratoryâ€™s experience. Genetics in Medicine, 2012, 14, 620-626.	2.4	39
90	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
91	Comprehensive One-Step Molecular Analyses of Mitochondrial Genome by Massively Parallel Sequencing. Clinical Chemistry, 2012, 58, 1322-1331.	3.2	144
92	Diagnostic approaches to apparent homozygosity. Genetics in Medicine, 2012, 14, 877-882.	2.4	38
93	SURF1-associated leigh syndrome: A case series and novel mutations. Human Mutation, 2012, 33, 1192-1200.	2.5	40
94	Application of Next Generation Sequencing to Molecular Diagnosis of Inherited Diseases. Topics in Current Chemistry, 2012, 336, 19-45.	4.0	43
95	Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE)-like phenotype: an expanded clinical spectrum of POLG1 mutations. Journal of Neurology, 2012, 259, 862-868.	3.6	112
96	Mitochondrial tRNA-serine (AGY) m.C12264T mutation causes severe multisystem disease with cataracts. Discovery Medicine, 2012, 13, 143-50.	0.5	4
97	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
98	Realâ€Time Quantitative PCR Analysis of Mitochondrial DNA Content. Current Protocols in Human Genetics, 2011, 68, Unit 19.7..	3.5	90
99	Analysis of Mitochondrial DNA Point Mutation Heteroplasmy by ARMS Quantitative PCR. Current Protocols in Human Genetics, 2011, 68, Unit 19.6..	3.5	25
100	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
101	Molecular characterization of CPS1 deletions by array CGH. Molecular Genetics and Metabolism, 2011, 102, 103-106.	1.1	11
102	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
103	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
104	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
105	Deoxyguanosine kinase deficiency presenting as neonatal hemochromatosis. Molecular Genetics and Metabolism, 2011, 103, 262-267.	1.1	33
106	Early-onset severe neuromuscular phenotype associated with compound heterozygosity for OPA1 mutations. Molecular Genetics and Metabolism, 2011, 103, 383-387.	1.1	71
107	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
108	Mitochondrial DNA Content Varies with Pathological Characteristics of Breast Cancer. Journal of Oncology, 2011, 2011, 1-10.	1.3	52

#	Article	IF	Citations
109	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
110	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
111	Mitochondrial DNA polymerase Â mutations: an ever expanding molecular and clinical spectrum. Journal of Medical Genetics, 2011, 48, 669-681.	3.2	140
112	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
113	Biochemical analysis of human POLG2 variants associated with mitochondrial disease. Human Molecular Genetics, 2011, 20, 3052-3066.	2.9	57
114	Novel POLG Splice Site Mutation and Optic Atrophy. Archives of Neurology, 2011, 68, 806-11.	4.5	29
115	POLG-related disorders. Neurology, 2011, 77, 1847-1852.	1.1	31
116	Mitochondrial dysfunction in human breast cancer cells and their transmitochondrial cybrids. Biochimica Et Biophysica Acta - Bioenergetics, 2010, 1797, 29-37.	1.0	77
117	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
118	Molecular genetics of mitochondrial disorders. Developmental Disabilities Research Reviews, 2010, 16, 154-162.	2.9	62
119	Exercise intolerance due to cytochrome <i>b</i> mutation. Muscle and Nerve, 2010, 42, 136-140.	2.2	19
120	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
121	Maternal systemic primary carnitine deficiency uncovered by newborn screening: Clinical, biochemical, and molecular aspects. Genetics in Medicine, 2010, 12, 19-24.	2.4	91
122	A novel c.592-4_c.592-3delTT mutation in DGUOK gene causes exon skipping. Mitochondrion, 2010, 10, 188-191.	3.4	13
123	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
124	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
125	MPV17-associated hepatocerebral mitochondrial DNA depletion syndrome: New patients and novel mutations. Molecular Genetics and Metabolism, 2010, 99, 300-308.	1.1	80
126	Current molecular diagnostic algorithm for mitochondrial disorders. Molecular Genetics and Metabolism, 2010, 100, 111-117.	1.1	66

#	Article	IF	Citations
127	Somatic mosaicism for PDHA1 mutation in a male with pyruvate dehydrogenase complex deficiency. Molecular Genetics and Metabolism, 2010, 100, 296-299.	1.1	13
128	Quantitative Evaluation of the Mitochondrial DNA Depletion Syndrome. Clinical Chemistry, 2010, 56, 1119-1127.	3.2	119
129	Sequence Homology at the Breakpoint and Clinical Phenotype of Mitochondrial DNA Deletion Syndromes. PLoS ONE, 2010, 5, e15687.	2.5	49
130	Mitochondrial Neurogastrointestinal Encephalopathy Due to Mutations in RRM2B. Archives of Neurology, 2009, 66, 1028-32.	4.5	103
131	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
132	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
133	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
134	Citrin deficiency, a perplexing global disorder. Molecular Genetics and Metabolism, 2009, 96, 44-49.	1.1	81
135	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
136	Mitochondrial disorder with OPA1 mutation lacking optic atrophy. Mitochondrion, 2009, 9, 279-281.	3.4	37
137	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
138	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
139	Mitochondrial DNA variant interactions modify breast cancer risk. Journal of Human Genetics, 2008, 53, 924-928.	2.3	49
140	Human mitochondrial transfer RNAs: Role of pathogenic mutation in disease. Muscle and Nerve, 2008, 37, 150-171.	2.2	68
141	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
142	Molecular and clinical genetics of mitochondrial diseases due toPOLGmutations. Human Mutation, 2008, 29, E150-E172.	2.5	256
143	Alpers syndrome with prominent white matter changes. Brain and Development, 2008, 30, 295-300.	1.1	17
144	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

#	Article	IF	Citations
145	Sensory ataxic neuropathy with ophthalmoparesis caused by POLG mutations. Neuromuscular Disorders, 2008, 18, 626-632.	0.6	71
146	The in-depth evaluation of suspected mitochondrial disease. Molecular Genetics and Metabolism, 2008, 94, 16-37.	1.1	320
147	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
148	Utility of Oligonucleotide Arrayâ€"Based Comparative Genomic Hybridization for Detection of Target Gene Deletions. Clinical Chemistry, 2008, 54, 1141-1148.	3.2	78
149	Mitochondrial immunofluorescence assay as an adjunctive tool in the diagnosis of mitochondrial myopathy. FASEB Journal, 2008, 22, 708.20.	0.5	0
150	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
151	Diagnostic challenges of mitochondrial DNA disorders. Mitochondrion, 2007, 7, 45-52.	3.4	75
152	Mutations in theMPV17 gene are responsible for rapidly progressive liver failure in infancy. Hepatology, 2007, 46, 1218-1227.	7.3	111
153	Pathogenic mitochondrial DNA mutations in protein-coding genes. Muscle and Nerve, 2007, 36, 279-293.	2.2	109
154	First prenatal exclusion of cystic fibrosis in East Asia. Pediatrics International, 2007, 49, 686-687.	0.5	0
155	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
156	AIB1 gene amplification and the instability of polyQ encoding sequence in breast cancer cell lines. BMC Cancer, 2006, 6, 111.	2.6	13
157	Exercise intolerance associated with a novel 8300t>C mutation in mitochondrial transfer RNAlys. Muscle and Nerve, 2006, 34, 437-443.	2.2	9
158	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
159	Molecular Analysis of Mitochondrial DNA Point Mutations by Polymerase Chain Reaction. , 2006, 336, 135-144.		10
160	Real-Time Quantitative Polymerase Chain Reaction Analysis of Mitochondrial DNA Point Mutation. , 2006, 335, 187-200.		11
161	Mitochondrial DNA analysis in clinical laboratory diagnostics. Clinica Chimica Acta, 2005, 354, 1-20.	1.1	83
162	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

#	Article	IF	Citations
163	Molecular genetics of common mitochondrial DNA disorders. Beijing Da Xue Xue Bao, 2005, 37, 26-31.	0.2	0
164	Clinical Spectrum, Morbidity, and Mortality in 113 Pediatric Patients With Mitochondrial Disease. Pediatrics, 2004, 114, 925-931.	2.1	431
165	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
166	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
167	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
168	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
169	Variable clinical manifestation of homoplasmic G14459A mitochondrial DNA mutation. American Journal of Medical Genetics Part A, 2004, 124A, 377-382.	2.4	87
170	In reply to: Proving pathogenicity?when evolution is not enough. American Journal of Medical Genetics Part A, 2004, 131A, 109-110.	2.4	0
171	Detection ofCFTR mutations using temporal temperature gradient gel electrophoresis. Electrophoresis, 2004, 25, 2593-2601.	2.4	15
172	Detection of mitochondrial DNA mutations using temporal temperature gradient gel electrophoresis. Electrophoresis, 2004, 25, 2602-2610.	2.4	32
173	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
174	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
175	Molecular analysis for mitochondrial DNA disorders. Mitochondrion, 2004, 4, 403-415.	3.4	38
176	The necessity of complete mutational analysis of an infertile couple before in vitro fertilization. Fertility and Sterility, 2004, 82, 947-949.	1.0	16
177	Somatic Mitochondrial DNA Mutations in Neurofibromatosis Type 1-Associated Tumors. Molecular Cancer Research, 2004, 2, 433-441.	3.4	45
178	Detection of mitochondrial DNA mutations in the tumor and cerebrospinal fluid of medulloblastoma patients. Cancer Research, 2003, 63, 3866-71.	0.9	67
179	Intergenerational transmission of pathogenic heteroplasmic mitochondrial DNA. Genetics in Medicine, 2002, 4, 78-83.	2.4	20
180	Comprehensive Scanning of the Entire Mitochondrial Genome for Mutations. Clinical Chemistry, 2002, 48, 1901-1912.	3.2	85

#	Article	IF	Citations
181	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
182	Comprehensive scanning of somatic mitochondrial DNA mutations in breast cancer. Cancer Research, 2002, 62, 972-6.	0.9	192
183	Comprehensive scanning of the entire mitochondrial genome for mutations. Clinical Chemistry, 2002, 48, 1901-12.	3.2	30
184	Recognition of mitochondrial DNA deletion syndrome with non-neuromuscular multisystemic manifestation. Genetics in Medicine, 2001, 3, 399-404.	2.4	28
185	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
186	Improved detection ofCFTR mutations in Southern California Hispanic CF patients. Human Mutation, 2001, 18, 296-307.	2.5	48
187	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
188	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
189	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
190	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
191	Detection of Mitochondrial DNA Mutations by Temporal Temperature Gradient Gel Electrophoresis. Clinical Chemistry, 1999, 45, 1162-1167.	3.2	82
192	Yield of mtDNA mutation analysis in 2,000 patients. , 1998, 77, 395-400.		84
193	Preparation and Validation of PCR-generated Positive Controls for Diagnostic Dot Blotting. Clinical Chemistry, 1998, 44, 1578-1579.	3.2	22
194	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
195	Yield of mtDNA mutation analysis in 2,000 patients. American Journal of Medical Genetics Part A, 1998, 77, 395-400.	2.4	1
196	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
197	Cyclic vomiting syndrome and mitochondrial DNA mutations. Lancet, The, 1997, 350, 1299-1300.	13.7	88
198	Direct detection of multiple point mutations in mitochondrial DNA. Clinical Chemistry, 1997, 43, 1857-1861.	3.2	84

#	Article	IF	Citations
199	Alternative, Noninvasive Tissues for Quantitative Screening of Mutant Mitochondrial DNA. Clinical Chemistry, 1997, 43, 1241-1243.	3.2	49
200	PRENATAL DIAGNOSIS OF GLYCOGEN STORAGE DISEASE TYPE 1a BY DIRECT MUTATION DETECTION. , 1996, 16, 105-108.		13
201	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
202	Diagnostic approaches to apparent homozygosity. , 0, .		1