Richard J Rodenburg

List of Publications by Year
in descending order

266 papers	13,985 citations	16451 64 h-index	31849 101 g-index
285 all docs	285 docs citations	285 times ranked	18079 citing authors

#	Article	IF	Citations
1	Gitelman-Like Syndrome Caused by Pathogenic Variants in mtDNA. Journal of the American Society of Nephrology: JASN, 2022, 33, 305-325.	6.1	26
2	Variants in Mitochondrial <scp>ATP</scp> Synthase Cause Variable Neurologic Phenotypes. Annals of Neurology, 2022, 91, 225-237.	5.3	12
3	Mitochondrial <scp>RNA</scp> processing defect caused by a <scp>SUPV3L1</scp> mutation in two siblings with a novel neurodegenerative syndrome. Journal of Inherited Metabolic Disease, 2022, 45, 292-307.	3.6	6
4	DTYMK is essential for genome integrity and neuronal survival. Acta Neuropathologica, 2022, 143, 245-262.	7.7	11
5	How to proceed after â€œnegativeâ€•exome: A review on genetic diagnostics, limitations, challenges, and emerging new multiomics techniques. Journal of Inherited Metabolic Disease, 2022, 45, 663-681.	3.6	20
6	RRM1 variants cause a mitochondrial DNA maintenance disorder via impaired de novo nucleotide synthesis. Journal of Clinical Investigation, 2022, 132, .	8.2	6
7	The decylTPP mitochondria-targeting moiety lowers electron transport chain supercomplex levels in primary human skin fibroblasts. Free Radical Biology and Medicine, 2022, 188, 434-446.	2.9	5
8	Chronic fluoxetine or ketamine treatment differentially affects brain energy homeostasis which is not exacerbated in mice with trait suboptimal mitochondrial function. European Journal of Neuroscience, 2021, 53, 2986-3001.	2.6	8
9	A novel variant in COX16 causes cytochrome c oxidase deficiency, severe fatal neonatal lactic acidosis, encephalopathy, cardiomyopathy, and liver dysfunction. Human Mutation, 2021, 42, 135-141.	2.5	4
10	Exome sequencing in paediatric patients with movement disorders. Orphanet Journal of Rare Diseases, 2021, 16, 32.	2.7	15
11	One mutation, three phenotypes: novel metabolic insights on MELAS, MIDD and myopathy caused by the m.3243Aâ€‰>â€‰G mutation. Metabolomics, 2021, 17, 10.	3.0	11
12	Loss of sdhb in zebrafish larvae recapitulates human paraganglioma characteristics. Endocrine-Related Cancer, 2021, 28, 65-77.	3.1	9
13	Long-term treated HIV infection is associated with platelet mitochondrial dysfunction. Scientific Reports, 2021, 11, 6246.	3.3	17
14	Severe Form of ÃŸIV-Spectrin Deficiency With Mitochondrial Dysfunction and Cardiomyopathyâ€”A Case Report. Frontiers in Neurology, 2021, 12, 643805.	2.4	4
15	Soluble adenylyl cyclase regulates the cytosolic NADH/NAD+ redox state and the bioenergetic switch between glycolysis and oxidative phosphorylation. Biochimica Et Biophysica Acta - Bioenergetics, 2021, 1862, 148367.	1.0	12
16	Human <scp>d</scp>â€lactate dehydrogenase deficiency by LDHD mutation in a patient with neurological manifestations and mitochondrial complex IV deficiency. JIMD Reports, 2021, 60, 15-22.	1.5	6
17	A MT-TL1 variant identified by whole exome sequencing in an individual with intellectual disability, epilepsy, and spastic tetraparesis. European Journal of Human Genetics, 2021, 29, 1359-1368.	2.8	7
18	Effect of neuropsychiatric medications on mitochondrial function: For better or for worse. Neuroscience and Biobehavioral Reviews, 2021, 127, 555-571.	6.1	15

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19	Moderate Intensity Exercise Training Improves Skeletal Muscle Performance inÂSymptomatic and Asymptomatic StatinÂUsers. Journal of the American College of Cardiology, 2021, 78, 2023-2037.	2.8	13
20	Characterization of a Novel Splicing Variant in Acylglycerol Kinase (AGK) Associated with Fatal Sengers Syndrome. International Journal of Molecular Sciences, 2021, 22, 13484.	4.1	6
21	Bi-Allelic UQCRFS1 Variants Are Associated with Mitochondrial Complex III Deficiency, Cardiomyopathy, and Alopecia Totalis. American Journal of Human Genetics, 2020, 106, 102-111.	6.2	36
22	Clinical, morphological and genetic characterization of Brody disease: an international study of 40 patients. Brain, 2020, 143, 452-466.	7.6	22
23	Identification of a Novel Variant in EARS2 Associated with a Severe Clinical Phenotype Expands the Clinical Spectrum of LTBL. Genes, 2020, 11, 1028.	2.4	2
24	Delineation of molecular findings by whole-exome sequencing for suspected cases of paediatric-onset mitochondrial diseases in the Southern Chinese population. Human Genomics, 2020, 14, 28.	2.9	11
25	Impaired mitochondrial complex I function as a candidate driver in the biological stress response and a concomitant stress-induced brain metabolic reprogramming in male mice. Translational Psychiatry, 2020, 10, 176.	4.8	33
26	Mutations in the Vâ€ATPase Assembly Factor VMA21 Cause a Congenital Disorder of Glycosylation With Autophagic Liver Disease. Hepatology, 2020, 72, 1968-1986.	7.3	32
27	Variants in NGLY1 lead to intellectual disability, myoclonus epilepsy, sensorimotor axonal polyneuropathy and mitochondrial dysfunction. Clinical Genetics, 2020, 97, 556-566.	2.0	19
28	TMEM70 functions in the assembly of complexes I and V. Biochimica Et Biophysica Acta - Bioenergetics, 2020, 1861, 148202.	1.0	31
29	Homozygous damaging SOD2 variant causes lethal neonatal dilated cardiomyopathy. Journal of Medical Genetics, 2020, 57, 23-30.	3.2	16
30	NDUFS4 deletion triggers loss of NDUFA12 in Ndufs4 mice and Leigh syndrome patients: A stabilizing role for NDUFAF2. Biochimica Et Biophysica Acta - Bioenergetics, 2020, 1861, 148213.	1.0	25
31	m.3243A > G-Induced Mitochondrial Dysfunction Impairs Human Neuronal Development and Reduces Neuronal Network Activity and Synchronicity. Cell Reports, 2020, 31, 107538.	6.4	56
32	KBTBD13 is an actin-binding protein that modulates muscle kinetics. Journal of Clinical Investigation, 2020, 130, 754-767.	8.2	25
33	Primary coenzyme Q10 deficiency-7: expanded phenotypic spectrum and a founder mutation in southern Chinese. Npj Genomic Medicine, 2019, 4, 18.	3.8	29
34	Bi-allelic GOT2 Mutations Cause a Treatable Malate-Aspartate Shuttle-Related Encephalopathy. American Journal of Human Genetics, 2019, 105, 534-548.	6.2	46
35	ARX-associated infantile epileptic-dyskinetic encephalopathy with responsiveness to valproate for controlling seizures and reduced activity of muscle mitochondrial complex IV. Brain and Development, 2019, 41, 883-887.	1.1	10
36	Loss of Bardet-Biedl syndrome proteins causes synaptic aberrations in principal neurons. PLoS Biology, 2019, 17, e3000414.	5.6	17

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37	Identification and Characterization of New Variants in FOXRED1 Gene Expands the Clinical Spectrum Associated with Mitochondrial Complex I Deficiency. Journal of Clinical Medicine, 2019, 8, 1262.	2.4	5
38	A fatal case of COQ7â€associated primary coenzyme Q₁₀ deficiency. JIMD Reports, 2019, 47, 23-29.	1.5	30
39	Skeletal muscle toxicity associated with tyrosine kinase inhibitor therapy in patients with chronic myeloid leukemia. Leukemia, 2019, 33, 2116-2120.	7.2	23
40	Panel-Based Nuclear and Mitochondrial Next-Generation Sequencing Outcomes of an Ethnically Diverse Pediatric Patient Cohort with Mitochondrial Disease. Journal of Molecular Diagnostics, 2019, 21, 503-513.	2.8	12
41	A Drosophila Mitochondrial Complex I Deficiency Phenotype Array. Frontiers in Genetics, 2019, 10, 245.	2.3	14
42	Biallelic variants in LARS2 and KARS cause deafness and (ovario)leukodystrophy. Neurology, 2019, 92, e1225-e1237.	1.1	32
43	Intraâ€patient variability of heteroplasmy levels in urinary epithelial cells in carriers of the m.3243A>G mutation. Molecular Genetics & Genomic Medicine, 2019, 7, e00523.	1.2	8
44	Mutated SUCLG1 causes mislocalization of SUCLG2 protein, morphological alterations of mitochondria and an early-onset severe neurometabolic disorder. Molecular Genetics and Metabolism, 2019, 126, 43-52.	1.1	20
45	Diverse phenotype in patients with complex I deficiency due to mutations in NDUFB11. European Journal of Medical Genetics, 2019, 62, 103572.	1.3	22
46	A urinary biosignature for mitochondrial myopathy, encephalopathy, lactic acidosis and stroke like episodes (MELAS). Mitochondrion, 2019, 45, 38-45.	3.4	16
47	Investigating the cardiac pathology of SCO2â€mediated hypertrophic cardiomyopathy using patients induced pluripotent stem cellâ€“derived cardiomyocytes. Journal of Cellular and Molecular Medicine, 2018, 22, 913-925.	3.6	27
48	MT-ND5 Mutation Exhibits Highly Variable Neurological Manifestations at Low Mutant Load. EBioMedicine, 2018, 30, 86-93.	6.1	47
49	Iron deficiency impairs contractility of human cardiomyocytes through decreased mitochondrial function. European Journal of Heart Failure, 2018, 20, 910-919.	7.1	225
50	QIL1-dependent assembly of MICOS complexâ€“lethal mutation in C19ORF70 resulting in liver disease and severe neurological retardation. Journal of Human Genetics, 2018, 63, 707-716.	2.3	29
51	A Heterozygous NDUFV1 Variant Aggravates Mitochondrial Complex I Deficiency in a Family with a Homoplasmic ND1 Variant. Journal of Pediatrics, 2018, 196, 309-313.e3.	1.8	13
52	The functional genomics laboratory: functional validation of genetic variants. Journal of Inherited Metabolic Disease, 2018, 41, 297-307.	3.6	48
53	Compound heterozygous SPATA5 variants in four families and functional studies of SPATA5 deficiency. European Journal of Human Genetics, 2018, 26, 407-419.	2.8	29
54	Mutations in SELENBP1, encoding a novel human methanethiol oxidase, cause extraoral halitosis. Nature Genetics, 2018, 50, 120-129.	21.4	86

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55	Feeding difficulties, a key feature of the Drosophila NDUFS4 mitochondrial disease model. DMM Disease Models and Mechanisms, 2018, 11, .	2.4	18
56	A family segregating lethal neonatal coenzyme Q₁₀ deficiency caused by mutations in COQ9. Journal of Inherited Metabolic Disease, 2018, 41, 719-729.	3.6	30
57	Bi-allelic Mutations in the Mitochondrial Ribosomal Protein MRPS2 Cause Sensorineural Hearing Loss, Hypoglycemia, and Multiple OXPHOS Complex Deficiencies. American Journal of Human Genetics, 2018, 102, 685-695.	6.2	61
58	Mining for mitochondrial mechanisms: Linking known syndromes to mitochondrial function. Clinical Genetics, 2018, 93, 943-951.	2.0	7
59	Statins Affect Skeletal Muscle Performance: Evidence for Disturbances in Energy Metabolism. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 75-84.	3.6	44
60	The genotypic and phenotypic spectrum of MTO1 deficiency. Molecular Genetics and Metabolism, 2018, 123, 28-42.	1.1	24
61	Pathogenic variants in glutamyl-tRNAGln amidotransferase subunits cause a lethal mitochondrial cardiomyopathy disorder. Nature Communications, 2018, 9, 4065.	12.8	44
62	Age-Dependent Decrease of Mitochondrial Complex II Activity in a Familial Mouse Model for Alzheimerâ€™s Disease. Journal of Alzheimer's Disease, 2018, 66, 75-82.	2.6	13
63	Coenzyme Q10 deficiency due to a COQ4 gene defect causes childhood-onset spinocerebellar ataxia and stroke-like episodes. Molecular Genetics and Metabolism Reports, 2018, 17, 19-21.	1.1	25
64	Effectiveness of whole exome sequencing in unsolved patients with a clinical suspicion of a mitochondrial disorder in Estonia. Molecular Genetics and Metabolism Reports, 2018, 15, 80-89.	1.1	37
65	Integrating glycomics and genomics uncovers SLC10A7 as essential factor for bone mineralization by regulating post-Golgi protein transport and glycosylation. Human Molecular Genetics, 2018, 27, 3029-3045.	2.9	37
66	Novel homozygous PCK1 mutation causing cytosolic phosphoenolpyruvate carboxykinase deficiency presenting as childhood hypoglycemia, an abnormal pattern of urine metabolites and liver dysfunction. Molecular Genetics and Metabolism, 2017, 120, 337-341.	1.1	29
67	Mutation in mitochondrial complex IV subunit COX5A causes pulmonary arterial hypertension, lactic acidemia, and failure to thrive. Human Mutation, 2017, 38, 692-703.	2.5	32
68	Mild orotic aciduria in UMPS heterozygotes: a metabolic finding without clinical consequences. Journal of Inherited Metabolic Disease, 2017, 40, 423-431.	3.6	14
69	Microbial stimulation of different Toll-like receptor signalling pathways induces diverse metabolic programmes in human monocytes. Nature Microbiology, 2017, 2, 16246.	13.3	228
70	Mutations in mitochondrial complex I assembly factor NDUFAF3 cause Leigh syndrome. Molecular Genetics and Metabolism, 2017, 120, 243-246.	1.1	21
71	4â€Hydroxybenzoic acid restores CoQ₁₀ biosynthesis in human <scp>COQ</scp>2 deficiency. Annals of Clinical and Translational Neurology, 2017, 4, 902-908.	3.7	25
72	NDUFAF4 variants are associated with Leigh syndrome and cause a specific mitochondrial complex I assembly defect. European Journal of Human Genetics, 2017, 25, 1273-1277.	2.8	25

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73	Biallelic variants inWARS2encoding mitochondrial tryptophanyl-tRNA synthase in six individuals with mitochondrial encephalopathy. Human Mutation, 2017, 38, 1786-1795.	2.5	24
74	Early and lethal neurodegeneration with myasthenic and myopathic features. Neurology, 2017, 89, 657-664.	1.1	20
75	Biallelic Mutations in LIPT2 Cause a Mitochondrial Lipoylation Defect Associated with Severe Neonatal Encephalopathy. American Journal of Human Genetics, 2017, 101, 283-290.	6.2	55
76	Recessive mutation in EXOSC3 associates with mitochondrial dysfunction and pontocerebellar hypoplasia. Mitochondrion, 2017, 37, 46-54.	3.4	26
77	Modulation of oxidative phosphorylation and redox homeostasis in mitochondrial NDUFS4 deficiency via mesenchymal stem cells. Stem Cell Research and Therapy, 2017, 8, 150.	5.5	26
78	A lethal neonatal phenotype of mitochondrial shortâ€chain enoylâ€‹scp›CoA‹/scp› hydrataseâ€1 deficiency. Clinical Genetics, 2017, 91, 629-633.	2.0	26
79	Detection of clinically relevant copy-number variants by exome sequencing in a large cohort of genetic disorders. Genetics in Medicine, 2017, 19, 667-675.	2.4	143
80	Coenzyme Q deficiency causes impairment of the sulfide oxidation pathway. EMBO Molecular Medicine, 2017, 9, 96-111.	6.9	61
81	Enzyme Diagnostics in a Changing World of Exome Sequencing and Newborn Screening as Exemplified for Peroxisomal, Mitochondrial, and Lysosomal Disorders. , 2017, , 461-487.		0
82	Mitochondrial complex I dysfunction and altered NAD(P)H kinetics in rat myocardium in cardiac right ventricular hypertrophy and failure. Cardiovascular Research, 2016, 111, 362-372.	3.8	42
83	Mutations in Complex I Assembly Factor TMEM126B Result in Muscle Weakness and Isolated Complex I Deficiency. American Journal of Human Genetics, 2016, 99, 208-216.	6.2	51
84	MIPEP recessive variants cause a syndrome of left ventricular non-compaction, hypotonia, and infantile death. Genome Medicine, 2016, 8, 106.	8.2	43
85	Mitochondrial disorders in children: toward development of smallâ€molecule treatment strategies. EMBO Molecular Medicine, 2016, 8, 311-327.	6.9	86
86	Three families with â€˜de novoâ€™ m.3243A>G mutation. BBA Clinical, 2016, 6, 19-24.	4.1	22
87	A novel mutation in FBXL4 in a Norwegian child with encephalomyopathic mitochondrial DNA depletion syndrome 13. European Journal of Medical Genetics, 2016, 59, 342-346.	1.3	16
88	RARS2 Mutations: Is Pontocerebellar Hypoplasia Type 6 a Mitochondrial Encephalopathy?. JIMD Reports, 2016, 33, 87-92.	1.5	14
89	Recurrent De Novo Dominant Mutations in SLC25A4 Cause Severe Early-Onset Mitochondrial Disease and Loss of Mitochondrial DNA Copy Number. American Journal of Human Genetics, 2016, 99, 860-876.	6.2	93
90	Leigh-Like Syndrome Due to Homoplasmic m.8993T>G Variant with Hypocitrullinemia and Unusual Biochemical Features Suggestive of Multiple Carboxylase Deficiency (MCD). JIMD Reports, 2016, 33, 99-107.	1.5	17

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91	Sudden Cardiac Death Due to Deficiency of the Mitochondrial Inorganic Pyrophosphatase PPA2. American Journal of Human Genetics, 2016, 99, 674-682.	6.2	48
92	ATP6AP1 deficiency causes an immunodeficiency with hepatopathy, cognitive impairment and abnormal protein glycosylation. Nature Communications, 2016, 7, 11600.	12.8	110
93	Bilateral Vestibulopathy Aggravates Balance and Gait Disturbances in Sensory Ataxic Neuropathy, Dysarthria, and Ophthalmoparesis: A Case Report. Journal of Clinical Neuromuscular Disease, 2016, 18, 34-36.	0.7	3
94	Primary skeletal muscle myoblasts from chronic heart failure patients exhibit loss of anti-inflammatory and proliferative activity. BMC Cardiovascular Disorders, 2016, 16, 107.	1.7	11
95	Quality of life, fatigue and mental health in patients with the m.3243Aâ€‰>â€‰G mutation and its correlates with genetic characteristics and disease manifestation. Orphanet Journal of Rare Diseases, 2016, 11, 25.	2.7	14
96	Cytosolic phosphoenolpyruvate carboxykinase deficiency presenting with acute liver failure following gastroenteritis. Molecular Genetics and Metabolism, 2016, 118, 21-27.	1.1	23
97	TMEM199 Deficiency Is a Disorder of Golgi Homeostasis Characterized by Elevated Aminotransferases, Alkaline Phosphatase, and Cholesterol and Abnormal Glycosylation. American Journal of Human Genetics, 2016, 98, 322-330.	6.2	73
98	Mitochondrial complex I-linked disease. Biochimica Et Biophysica Acta - Bioenergetics, 2016, 1857, 938-945.	1.0	137
99	Increased mitochondrial ATP production capacity in brain of healthy mice and a mouse model of isolated complex I deficiency after isoflurane anesthesia. Journal of Inherited Metabolic Disease, 2016, 39, 59-65.	3.6	10
100	Fatal neonatal encephalopathy and lactic acidosis caused by a homozygous loss-of-function variant in COQ9. European Journal of Human Genetics, 2016, 24, 450-454.	2.8	45
101	Deficiency of <scp>ECHS</scp>1 causes mitochondrial encephalopathy with cardiac involvement. Annals of Clinical and Translational Neurology, 2015, 2, 492-509.	3.7	90
102	Quantifying small molecule phenotypic effects using mitochondrial morpho-functional fingerprinting and machine learning. Scientific Reports, 2015, 5, 8035.	3.3	36
103	Novel, Compound Heterozygous, Single-Nucleotide Variants inMARS2Associated with Developmental Delay, Poor Growth, and Sensorineural Hearing Loss. Human Mutation, 2015, 36, 587-592.	2.5	29
104	SDHA mutations causing a multisystem mitochondrial disease: novel mutations and genetic overlap with hereditary tumors. European Journal of Human Genetics, 2015, 23, 202-209.	2.8	71
105	Atypical Clinical Presentations of TAZ Mutations: An Underdiagnosed Cause of Growth Retardation?. JIMD Reports, 2015, 29, 89-93.	1.5	8
106	Mitochondrial Disease Sequence Data Resource (MSeqDR): A global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities. Molecular Genetics and Metabolism, 2015, 114, 388-396.	1.1	76
107	A novel mitochondrial DNA m.7507A>G mutation is only pathogenic at high levels of heteroplasmy. Neuromuscular Disorders, 2015, 25, 262-267.	0.6	9
108	The spectrum of pyruvate oxidation defects in the diagnosis of mitochondrial disorders. Journal of Inherited Metabolic Disease, 2015, 38, 391-403.	3.6	44

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109	Mitochondrial dysfunction in primary human fibroblasts triggers an adaptive cell survival program that requires AMPK-Î±. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2015, 1852, 529-540.	3.8	40
110	CLPB Mutations Cause 3-Methylglutaconic Aciduria, Progressive Brain Atrophy, Intellectual Disability, Congenital Neutropenia, Cataracts, Movement Disorder. American Journal of Human Genetics, 2015, 96, 245-257.	6.2	111
111	Skeletal muscle mitochondria of NDUFS4âˆ’/âˆ’ mice display normal maximal pyruvate oxidation and ATP production. Biochimica Et Biophysica Acta - Bioenergetics, 2015, 1847, 526-533.	1.0	21
112	High prevalence of complementary and alternative medicine use in patients with genetically proven mitochondrial disorders. Journal of Inherited Metabolic Disease, 2015, 38, 477-482.	3.6	5
113	TRMT5 Mutations Cause a Defect in Post-transcriptional Modification of Mitochondrial tRNA Associated with Multiple Respiratory-Chain Deficiencies. American Journal of Human Genetics, 2015, 97, 319-328.	6.2	83
114	MRPL44 mutations cause a slowly progressive multisystem disease with childhood-onset hypertrophic cardiomyopathy. Neurogenetics, 2015, 16, 319-323.	1.4	44
115	Whole exome sequencing of suspected mitochondrial patients in clinical practice. Journal of Inherited Metabolic Disease, 2015, 38, 437-443.	3.6	186
116	SUCLA2 Deficiency: A Deafness-Dystonia Syndrome with Distinctive Metabolic Findings (Report of a) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	1.5	8
117	Muscle pain, fatigue and night hypothermia in association with mitochondrial dysfunction. Journal of Pediatric Neurology, 2015, 07, 345-350.	0.2	0
118	MRPS22 mutation causes fatal neonatal lactic acidosis with brain and heart abnormalities. Neurogenetics, 2015, 16, 237-240.	1.4	34
119	Statin-Induced Myopathy Is Associated with Mitochondrial Complex III Inhibition. Cell Metabolism, 2015, 22, 399-407.	16.2	180
120	Mutations inCOA6cause CytochromecOxidase Deficiency and Neonatal Hypertrophic Cardiomyopathy. Human Mutation, 2015, 36, 34-38.	2.5	72
121	New approaches to diagnosing mitochondrial abnormalities: Taking the next step. Journal of Pediatric Biochemistry, 2015, 02, 205-212.	0.2	0
122	Serum FGF21 levels in adult m.3243A>G carriers. Neurology, 2014, 83, 125-133.	1.1	33
123	Mutations in RARS cause hypomyelination. Annals of Neurology, 2014, 76, 134-139.	5.3	77
124	Phenylbutyrate increases pyruvate dehydrogenase complex activity in cells harboring a variety of defects. Annals of Clinical and Translational Neurology, 2014, 1, 462-470.	3.7	15
125	Mutations in APOPT1, Encoding a Mitochondrial Protein, Cause Cavitating Leukoencephalopathy with Cytochrome c Oxidase Deficiency. American Journal of Human Genetics, 2014, 95, 315-325.	6.2	64
126	The role of the mitochondrial ribosome in human disease: searching for mutations in 12S mitochondrial rRNA with high disruptive potential. Human Molecular Genetics, 2014, 23, 949-967.	2.9	35

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127	A mutation in the human CBP4 ortholog UQCC3 impairs complex III assembly, activity and cytochrome b stability. Human Molecular Genetics, 2014, 23, 6356-6365.	2.9	69
128	A guide to diagnosis and treatment of Leigh syndrome. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 257-265.	1.9	197
129	Phenotypic spectrum of eleven patients and five novel MTFMT mutations identified by exome sequencing and candidate gene screening. Molecular Genetics and Metabolism, 2014, 111, 342-352.	1.1	65
130	The role of mitochondrial OXPHOS dysfunction in the development of neurologic diseases. Neurobiology of Disease, 2013, 51, 27-34.	4.4	75
131	3â€Methylglutaconic aciduriaâ€"lessons from 50 genes and 977 patients. Journal of Inherited Metabolic Disease, 2013, 36, 913-921.	3.6	74
132	A Post-Hoc Comparison of the Utility of Sanger Sequencing and Exome Sequencing for the Diagnosis of Heterogeneous Diseases. Human Mutation, 2013, 34, 1721-1726.	2.5	303
133	A multi-center comparison of diagnostic methods for the biochemical evaluation of suspected mitochondrial disorders. Mitochondrion, 2013, 13, 36-43.	3.4	23
134	A novel mutation in COQ2 leading to fatal infantile multisystem disease. Journal of the Neurological Sciences, 2013, 326, 24-28.	0.6	45
135	Disturbed energy metabolism and muscular dystrophy caused by pure creatine deficiency are reversible by creatine intake. Journal of Physiology, 2013, 591, 571-592.	2.9	79
136	Leigh syndrome associated with mitochondrial complex I deficiency due to novel mutations In NDUFV1 and NDUFS2. Gene, 2013, 516, 162-167.	2.2	41
137	Socio-emotional Problems in Children with CDG. JIMD Reports, 2013, 11, 139-148.	1.5	3
138	A complex V ATP5A1 defect causes fatal neonatal mitochondrial encephalopathy. Brain, 2013, 136, 1544-1554.	7.6	80
139	NUBPL mutations in patients with complex I deficiency and a distinct MRI pattern. Neurology, 2013, 80, 1577-1583.	1.1	80
140	Mutations in the UQCC1-Interacting Protein, UQCC2, Cause Human Complex III Deficiency Associated with Perturbed Cytochrome b Protein Expression. PLoS Genetics, 2013, 9, e1004034.	3.5	96
141	A mutation in the FAM36A gene, the human ortholog of COX20, impairs cytochrome c oxidase assembly and is associated with ataxia and muscle hypotonia. Human Molecular Genetics, 2013, 22, 656-667.	2.9	75
142	Genotype-Specific Abnormalities in Mitochondrial Function Associate with Distinct Profiles of Energy Metabolism and Catecholamine Content in Pheochromocytoma and Paraganglioma. Clinical Cancer Research, 2013, 19, 3787-3795.	7.0	53
143	CEP89 is required for mitochondrial metabolism and neuronal function in man and fly. Human Molecular Genetics, 2013, 22, 3138-3151.	2.9	38
144	New Findings in a Global Approach to Dissect the Whole Phenotype of PLA2G6 Gene Mutations. PLoS ONE, 2013, 8, e76831.	2.5	42

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145	Analysis of 953 Human Proteins from a Mitochondrial HEK293 Fraction by Complexome Profiling. PLoS ONE, 2013, 8, e68340.	2.5	51
146	Gene identification in the congenital disorders of glycosylation type I by whole-exome sequencing. Human Molecular Genetics, 2012, 21, 4151-4161.	2.9	147
147	Fluorescence imaging of mitochondria in cultured skin fibroblasts: a useful method for the detection of oxidative phosphorylation defects. Pediatric Research, 2012, 72, 232-240.	2.3	16
148	Inheritance of the m.3243A>G mutation. JIMD Reports, 2012, 8, 47-50.	1.5	16
149	Mutations in the phospholipid remodeling gene SERAC1 impair mitochondrial function and intracellular cholesterol trafficking and cause dystonia and deafness. Nature Genetics, 2012, 44, 797-802.	21.4	175
150	C7orf30 specifically associates with the large subunit of the mitochondrial ribosome and is involved in translation. Nucleic Acids Research, 2012, 40, 4040-4051.	14.5	46
151	Novel Infantile-Onset Leukoencephalopathy With High Lactate Level and Slow Improvement. Archives of Neurology, 2012, 69, 718-22.	4.5	9
152	Clinical features and heteroplasmy in blood, urine and saliva in 34 Dutch families carrying the m.3243A > G mutation. Journal of Inherited Metabolic Disease, 2012, 35, 1059-1069.	3.6	120
153	Natural disease course and genotypeâ€phenotype correlations in Complex I deficiency caused by nuclear gene defects: what we learned from 130 cases. Journal of Inherited Metabolic Disease, 2012, 35, 737-747.	3.6	112
154	Mitochondrial DNA m.3242GÂ>ÂA mutation, an under diagnosed cause of hypertrophic cardiomyopathy and renal tubular dysfunction?. European Journal of Medical Genetics, 2012, 55, 552-556.	1.3	27
155	Metabolic consequences of NDUFS4 gene deletion in immortalized mouse embryonic fibroblasts. Biochimica Et Biophysica Acta - Bioenergetics, 2012, 1817, 1925-1936.	1.0	60
156	Transcriptional changes in OXPHOS complex I deficiency are related to anti-oxidant pathways and could explain the disturbed calcium homeostasis. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 1161-1168.	3.8	30
157	A catalytic defect in mitochondrial respiratory chain complex I due to a mutation in NDUFS2 in a patient with Leigh syndrome. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 168-175.	3.8	26
158	Trolox-Sensitive Reactive Oxygen Species Regulate Mitochondrial Morphology, Oxidative Phosphorylation and Cytosolic Calcium Handling in Healthy Cells. Antioxidants and Redox Signaling, 2012, 17, 1657-1669.	5.4	63
159	A Diagnostic Algorithm for Mitochondrial Disorders in Estonian Children. Molecular Syndromology, 2012, 3, 113-119.	0.8	5
160	Molecular base of biochemical complex I deficiency. Mitochondrion, 2012, 12, 520-532.	3.4	34
161	Impaired ubiquitinâ€proteasomeâ€mediated <scp>PGC</scp>â€1Î± protein turnover and induced mitochondrial biogenesis secondary to complexâ€<scp>I</scp> deficiency. Proteomics, 2012, 12, 1349-1362.	2.2	8
162	Lack of the Mitochondrial Protein Acylglycerol Kinase Causes Sengers Syndrome. American Journal of Human Genetics, 2012, 90, 314-320.	6.2	192

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