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	Mitochondrial ATP synthase: architecture, function and pathology. Journal of Inherited Metabolic Disease, 2012, 35, 211-225.	3.6	412
2	Leigh Syndrome with Nephropathy and CoQ10 Deficiency Due to decaprenyl diphosphate synthase subunit 2 (PDSS2) Mutations. American Journal of Human Genetics, 2006, 79, 1125-1129.	6.2	359
3	Spectrophotometric Assay for Complex I of the Respiratory Chain in Tissue Samples and Cultured Fibroblasts. Clinical Chemistry, 2007, 53, 729-734.	3.2	340
4	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
5	Mitochondrial complex I deficiency: from organelle dysfunction to clinical disease. Brain, 2008, 132, 833-842.	7.6	270
6	Overexpression of Akt converts radial growth melanoma to vertical growth melanoma. Journal of Clinical Investigation, 2007, 117, 719-729.	8.2	246
7	Mitochondrial disease criteria. Neurology, 2006, 67, 1823-1826.	1.1	243
8	Microbial stimulation of different Toll-like receptor signalling pathways induces diverse metabolic programmes in human monocytes. Nature Microbiology, 2017, 2, 16246.	13.3	228
9	Iron deficiency impairs contractility of human cardiomyocytes through decreased mitochondrial function. European Journal of Heart Failure, 2018, 20, 910-919.	7.1	225
10	A guide to diagnosis and treatment of Leigh syndrome. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 257-265.	1.9	197
11	Lack of the Mitochondrial Protein Acylglycerol Kinase Causes Sengers Syndrome. American Journal of Human Genetics, 2012, 90, 314-320.	6.2	192
12	Whole exome sequencing of suspected mitochondrial patients in clinical practice. Journal of Inherited Metabolic Disease, 2015, 38, 437-443.	3.6	186
13	SUCLA2 mutations are associated with mild methylmalonic aciduria, Leigh-like encephalomyopathy, dystonia and deafness. Brain, 2007, 130, 862-874.	7.6	180
14	Statin-Induced Myopathy Is Associated with Mitochondrial Complex III Inhibition. Cell Metabolism, 2015, 22, 399-407.	16.2	180
15	Enhanced number and activity of mitochondria in multiple sclerosis lesions. Journal of Pathology, 2009, 219, 193-204.	4.5	178
16	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
17	Acyl-CoA Dehydrogenase 9 Is Required for the Biogenesis of Oxidative Phosphorylation Complex I. Cell Metabolism, 2010, 12, 283-294.	16.2	172
18	Distinct Clinical Phenotypes Associated with a Mutation in the Mitochondrial Translation Elongation Factor EFTs. American Journal of Human Genetics, 2006, 79, 869-877.	6.2	169

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19	Depletion of PINK1 affects mitochondrial metabolism, calcium homeostasis and energy maintenance. Journal of Cell Science, 2011, 124, 1115-1125.	2.0	167
20	Biochemical diagnosis of mitochondrial disorders. Journal of Inherited Metabolic Disease, 2011, 34, 283-292.	3.6	165
21	Secondary mitochondrial dysfunction in propionic aciduria: a pathogenic role for endogenous mitochondrial toxins. Biochemical Journal, 2006, 398, 107-112.	3.7	163
22	Gene identification in the congenital disorders of glycosylation type I by whole-exome sequencing. Human Molecular Genetics, 2012, 21, 4151-4161.	2.9	147
23	Mutations in C12orf65 in Patients with Encephalomyopathy and a Mitochondrial Translation Defect. American Journal of Human Genetics, 2010, 87, 115-122.	6.2	144
24	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
25	Mitochondrial complex I-linked disease. Biochimica Et Biophysica Acta - Bioenergetics, 2016, 1857, 938-945.	1.0	137
26	A combination of proteomics, principal component analysis and transcriptomics is a powerful tool for the identification of biomarkers for macrophage maturation in the U937 cell line. Proteomics, 2004, 4, 1014-1028.	2.2	121
27	Cardiolipin and monolysocardiolipin analysis in fibroblasts, lymphocytes, and tissues using high-performance liquid chromatographyâ€“mass spectrometry as a diagnostic test for Barth syndrome. Analytical Biochemistry, 2009, 387, 230-237.	2.4	120
28	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
29	NDUFA2 Complex I Mutation Leads to Leigh Disease. American Journal of Human Genetics, 2008, 82, 1306-1315.	6.2	119
30	X-linkedNDUFA1gene mutations associated with mitochondrial encephalomyopathy. Annals of Neurology, 2007, 61, 73-83.	5.3	118
31	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
32	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
33	ATP6AP1 deficiency causes an immunodeficiency with hepatopathy, cognitive impairment and abnormal protein glycosylation. Nature Communications, 2016, 7, 11600.	12.8	110
34	Measurement of the Energy-Generating Capacity of Human Muscle Mitochondria: Diagnostic Procedure and Application to Human Pathology. Clinical Chemistry, 2006, 52, 860-871.	3.2	96
35	A novel mitochondrial ATP8 gene mutation in a patient with apical hypertrophic cardiomyopathy and neuropathy. Journal of Medical Genetics, 2007, 45, 129-133.	3.2	96
36	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

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37	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
38	The antiinflammatory drug sulfasalazine inhibits tumor necrosis factor Î± expression in macrophages by inducing apoptosis. Arthritis and Rheumatism, 2000, 43, 1941-1950.	6.7	92
39	Biochemical and genetic analysis of 3-methylglutaconic aciduria type IV: a diagnostic strategy. Brain, 2009, 132, 136-146.	7.6	90
40	Mutation in mitochondrial ribosomal protein MRPS22 leads to Cornelia de Lange-like phenotype, brain abnormalities and hypertrophic cardiomyopathy. European Journal of Human Genetics, 2011, 19, 394-399.	2.8	90
41	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
42	Mitochondrial disorders in children: toward development of smallâ€molecule treatment strategies. EMBO Molecular Medicine, 2016, 8, 311-327.	6.9	86
43	Mutations in SELENBP1, encoding a novel human methanethiol oxidase, cause extraoral halitosis. Nature Genetics, 2018, 50, 120-129.	21.4	86
44	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
45	NDUFA10 mutations cause complex I deficiency in a patient with Leigh disease. European Journal of Human Genetics, 2011, 19, 270-274.	2.8	82
46	LCâ€MS/MS as an alternative for SDSâ€PAGE in blue native analysis of protein complexes. Proteomics, 2009, 9, 4221-4228.	2.2	80
47	A complex V ATP5A1 defect causes fatal neonatal mitochondrial encephalopathy. Brain, 2013, 136, 1544-1554.	7.6	80
48	NUBPL mutations in patients with complex I deficiency and a distinct MRI pattern. Neurology, 2013, 80, 1577-1583.	1.1	80
49	Major depression in adolescent children consecutively diagnosed with mitochondrial disorder. Journal of Affective Disorders, 2009, 114, 327-332.	4.1	79
50	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
51	Development of an Androgen Reporter Gene Assay (AR-LUX) Utilizing a Human Cell Line with an Endogenously Regulated Androgen Receptor. Analytical Biochemistry, 2001, 298, 93-102.	2.4	78
52	Mutations in RARS cause hypomyelination. Annals of Neurology, 2014, 76, 134-139.	5.3	77
53	Mutated ND2 impairs mitochondrial complex I assembly and leads to Leigh Syndrome. Molecular Genetics and Metabolism, 2007, 90, 10-14.	1.1	76
54	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

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55	A Mutation in C2orf64 Causes Impaired Cytochrome c Oxidase Assembly and Mitochondrial Cardiomyopathy. American Journal of Human Genetics, 2011, 88, 488-493.	6.2	75
56	The role of mitochondrial OXPHOS dysfunction in the development of neurologic diseases. Neurobiology of Disease, 2013, 51, 27-34.	4.4	75
57	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
58	3â€Methylglutaconic aciduriaâ€"lessons from 50 genes and 977 patients. Journal of Inherited Metabolic Disease, 2013, 36, 913-921.	3.6	74
59	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
60	Unheated Cannabis sativa extracts and its major compound THC-acid have potential immuno-modulating properties not mediated by CB1 and CB2 receptor coupled pathways. International Immunopharmacology, 2006, 6, 656-665.	3.8	72
61	Mutations inCOA6cause CytochromecOxidase Deficiency and Neonatal Hypertrophic Cardiomyopathy. Human Mutation, 2015, 36, 34-38.	2.5	72
62	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
63	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
64	Investigation of the complex I assembly chaperones B17.2L and NDUFAF1 in a cohort of CI deficient patients. Molecular Genetics and Metabolism, 2007, 91, 176-182.	1.1	68
65	Clinical and biochemical characteristics in patients with a high mutant load of the mitochondrial T8993G/C mutations. American Journal of Medical Genetics, Part A, 2006, 140A, 863-868.	1.2	67
66	Depressive behaviour in children diagnosed with a mitochondrial disorder. Mitochondrion, 2010, 10, 528-533.	3.4	67
67	Characterization of anti-inflammatory compounds using transcriptomics, proteomics, and metabolomics in combination with multivariate data analysis. International Immunopharmacology, 2004, 4, 1499-1514.	3.8	66
68	Multiple oxidative phosphorylation deficiencies in severe childhood multi-system disorders due to polymerase gamma (POLG1) mutations. European Journal of Pediatrics, 2007, 166, 229-234.	2.7	66
69	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
70	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
71	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
72	Coenzyme Q deficiency causes impairment of the sulfide oxidation pathway. EMBO Molecular Medicine, 2017, 9, 96-111.	6.9	61

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73	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
74	Cell death: a trigger of autoimmunity?. BioEssays, 2000, 22, 627-636.	2.5	60
75	Metabolic consequences of NDUFS4 gene deletion in immortalized mouse embryonic fibroblasts. Biochimica Et Biophysica Acta - Bioenergetics, 2012, 1817, 1925-1936.	1.0	60
76	Association of 3-methylglutaconic aciduria with sensori-neural deafness, encephalopathy, and Leigh-like syndrome (MEGDEL association) in four patients with a disorder of the oxidative phosphorylation. Molecular Genetics and Metabolism, 2006, 88, 47-52.	1.1	59
77	Expression of macrophage-derived chemokine (MDC) mRNA in macrophages is enhanced by interleukin-1 Î², tumor necrosis factor Î±, and lipopolysaccharide. Journal of Leukocyte Biology, 1998, 63, 606-611.	3.3	58
78	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
79	The mitochondrial 13513G>A mutation is most frequent in Leigh syndrome combined with reduced complex I activity, optic atrophy and/or Wolffâ€“Parkinsonâ€“White. European Journal of Human Genetics, 2007, 15, 155-161.	2.8	55
80	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
81	Mitochondrial enzymes discriminate between mitochondrial disorders and chronic fatigue syndrome. Mitochondrion, 2011, 11, 735-738.	3.4	54
82	Respiratory chain complex I deficiency due to NDUFA12 mutations as a new cause of Leigh syndrome. Journal of Medical Genetics, 2011, 48, 737-740.	3.2	54
83	Novel mutations in the NDUFS1 gene cause low residual activities in human complex I deficiencies. Molecular Genetics and Metabolism, 2010, 100, 251-256.	1.1	53
84	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
85	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
86	Analysis of 953 Human Proteins from a Mitochondrial HEK293 Fraction by Complexome Profiling. PLoS ONE, 2013, 8, e68340.	2.5	51
87	Sudden Cardiac Death Due to Deficiency of the Mitochondrial Inorganic Pyrophosphatase PPA2. American Journal of Human Genetics, 2016, 99, 674-682.	6.2	48
88	The functional genomics laboratory: functional validation of genetic variants. Journal of Inherited Metabolic Disease, 2018, 41, 297-307.	3.6	48
89	MT-ND5 Mutation Exhibits Highly Variable Neurological Manifestations at Low Mutant Load. EBioMedicine, 2018, 30, 86-93.	6.1	47
90	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

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91	Bi-allelic GOT2 Mutations Cause a Treatable Malate-Aspartate Shuttle-Related Encephalopathy. American Journal of Human Genetics, 2019, 105, 534-548.	6.2	46
92	A novel mutation in COQ2 leading to fatal infantile multisystem disease. Journal of the Neurological Sciences, 2013, 326, 24-28.	0.6	45
93	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
94	Baculovirus complementation restores a novelNDUFAF2mutation causing complex I deficiency. Human Mutation, 2009, 30, E728-E736.	2.5	44
95	The spectrum of pyruvate oxidation defects in the diagnosis of mitochondrial disorders. Journal of Inherited Metabolic Disease, 2015, 38, 391-403.	3.6	44
96	MRPL44 mutations cause a slowly progressive multisystem disease with childhood-onset hypertrophic cardiomyopathy. Neurogenetics, 2015, 16, 319-323.	1.4	44
97	Statins Affect Skeletal Muscle Performance: Evidence for Disturbances in Energy Metabolism. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 75-84.	3.6	44
98	Pathogenic variants in glutamyl-tRNAGln amidotransferase subunits cause a lethal mitochondrial cardiomyopathy disorder. Nature Communications, 2018, 9, 4065.	12.8	44
99	Complex I disorders: Causes, mechanisms, and development of treatment strategies at the cellular level. Developmental Disabilities Research Reviews, 2010, 16, 175-182.	2.9	43
100	MIPEP recessive variants cause a syndrome of left ventricular non-compaction, hypotonia, and infantile death. Genome Medicine, 2016, 8, 106.	8.2	43
101	Mitochondrial dysfunction and organic aciduria in five patients carrying mutations in the Ras-MAPK pathway. European Journal of Human Genetics, 2011, 19, 138-144.	2.8	42
102	Mutation in subdomain G' of mitochondrial elongation factor G1 is associated with combined OXPHOS deficiency in fibroblasts but not in muscle. European Journal of Human Genetics, 2011, 19, 275-279.	2.8	42
103	New Findings in a Global Approach to Dissect the Whole Phenotype of PLA2G6 Gene Mutations. PLoS ONE, 2013, 8, e76831.	2.5	42
104	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
105	The liver-specific promoter of the human insulin-like growth factor II gene is activated by CCAAT/enhancer binding protein (C/EBP). Nucleic Acids Research, 1992, 20, 3099-3104.	14.5	41
106	Metabolic capacity of the diaphragm in patients with COPD. Respiratory Medicine, 2006, 100, 1064-1071.	2.9	41
107	Leigh syndrome associated with mitochondrial complex I deficiency due to novel mutations In NDUFV1 and NDUFS2. Gene, 2013, 516, 162-167.	2.2	41
108	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

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109	Transcriptional and Post-Transcriptional Regulation of the Human IGF-II Gene Expression. Advances in Experimental Medicine and Biology, 1994, 343, 63-71.	1.6	40
110	Endotoxin-induced liver damage in rats is minimized by ï¿½ 2 -adrenoceptor stimulation. Inflammation Research, 2004, 53, 93-99.	4.0	39
111	Restoration of complex V deficiency caused by a novel deletion in the human TMEM70 gene normalizes mitochondrial morphology. Mitochondrion, 2011, 11, 954-963.	3.4	39
112	CEP89 is required for mitochondrial metabolism and neuronal function in man and fly. Human Molecular Genetics, 2013, 22, 3138-3151.	2.9	38
113	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
114	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
115	Biochemical examination of fibroblasts in the diagnosis and research of oxidative phosphorylation (OXPHOS) defects. Mitochondrion, 2004, 4, 395-401.	3.4	36
116	Quantifying small molecule phenotypic effects using mitochondrial morpho-functional fingerprinting and machine learning. Scientific Reports, 2015, 5, 8035.	3.3	36
117	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
118	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
119	Dietary intervention and oxidative phosphorylation capacity. Journal of Inherited Metabolic Disease, 2006, 29, 589-589.	3.6	34
120	Isolated deficiencies of OXPHOS complexes I and IV are identified accurately and quickly by simple enzyme activity immunocapture assays. Biochimica Et Biophysica Acta - Bioenergetics, 2009, 1787, 533-538.	1.0	34
121	Molecular base of biochemical complex I deficiency. Mitochondrion, 2012, 12, 520-532.	3.4	34
122	MRPS22 mutation causes fatal neonatal lactic acidosis with brain and heart abnormalities. Neurogenetics, 2015, 16, 237-240.	1.4	34
123	Leigh Disease with Brainstem Involvement in Complex I Deficiency due to Assembly Factor NDUFAF2 Defect. Neuropediatrics, 2010, 41, 30-34.	0.6	33
124	Serum FGF21 levels in adult m.3243A>G carriers. Neurology, 2014, 83, 125-133.	1.1	33
125	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
126	Mitochondrial Energy Production Correlates With the Age-Related BMI. Pediatric Research, 2009, 65, 103-108.	2.3	32

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127	Functional consequences of mitochondrial tRNATrp and tRNAArg mutations causing combined OXPHOS defects. European Journal of Human Genetics, 2010, 18, 324-329.	2.8	32
128	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
129	Biallelic variants in LARS2 and KARS cause deafness and (ovario)leukodystrophy. Neurology, 2019, 92, e1225-e1237.	1.1	32
130	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
131	Androgenic activity in surface water samples detected using the AR-LUX assay: indications for mixture effects. Environmental Toxicology and Pharmacology, 2005, 19, 263-272.	4.0	31
132	Beta-adrenergic receptor agonists induce the release of granulocyte chemotactic protein-2, oncostatin M, and vascular endothelial growth factor from macrophages. International Immunopharmacology, 2006, 6, 1-7.	3.8	31
133	TMEM70 functions in the assembly of complexes I and V. Biochimica Et Biophysica Acta - Bioenergetics, 2020, 1861, 148202.	1.0	31
134	Skeletal Muscle Ultrasonography in Children with a Dysfunction in the Oxidative Phosphorylation System. Neuropediatrics, 2006, 37, 142-147.	0.6	30
135	MR spectroscopy of the brain in Leigh syndrome. Brain and Development, 2008, 30, 579-583.	1.1	30
136	MR Spectroscopy and Serial Magnetic Resonance Imaging in a Patient with Mitochondrial Cystic Leukoencephalopathy due to Complex I Deficiency andNDUFV1Mutations and Mild Clinical Course. Neuropediatrics, 2008, 39, 172-175.	0.6	30
137	Contiguous gene deletion of ELOVL7, ERCC8 and NDUFAF2 in a patient with a fatal multisystem disorder. Human Molecular Genetics, 2009, 18, 3365-3374.	2.9	30
138	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
139	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
140	A fatal case of COQ7â€associated primary coenzyme Q₁₀ deficiency. JIMD Reports, 2019, 47, 23-29.	1.5	30
141	Early cardiac involvement in children carrying the A3243G mtDNA mutation. Acta Paediatrica, International Journal of Paediatrics, 2007, 96, 450-451.	1.5	29
142	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
143	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
144	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

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145	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
146	Primary coenzyme Q10 deficiency-7: expanded phenotypic spectrum and a founder mutation in southern Chinese. Npj Genomic Medicine, 2019, 4, 18.	3.8	29
147	A Functional Sp1 Binding Site Is Essential for the Activity of the Adult Liver-Specific Human Insulin-Like Growth Factor II Promoter. Molecular Endocrinology, 1997, 11, 237-250.	3.7	27
148	In Search of Secreted Protein Biomarkers for the Anti-inflammatory Effect of Î²2-Adrenergic Receptor Agonists:Â Application of DIGE Technology in Combination with Multivariate and Univariate Data Analysis Tools. Journal of Proteome Research, 2005, 4, 2015-2023.	3.7	27
149	Females with PDHA1 gene mutations: A diagnostic challenge. Mitochondrion, 2006, 6, 155-159.	3.4	27
150	A Novel Mutation in the SCO2 Gene in a Neonate With Early-Onset Cardioencephalomyopathy. Pediatric Neurology, 2010, 42, 227-230.	2.1	27
151	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
152	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
153	Coenzyme Q₁₀ is decreased in fibroblasts of patients with methylmalonic aciduria but not in mevalonic aciduria. Journal of Inherited Metabolic Disease, 2009, 32, 570-575.	3.6	26
154	High-Throughput Assay to Measure Oxygen Consumption in Digitonin-Permeabilized Cells of Patients with Mitochondrial Disorders. Clinical Chemistry, 2010, 56, 424-431.	3.2	26
155	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
156	Recessive mutation in EXOSC3 associates with mitochondrial dysfunction and pontocerebellar hypoplasia. Mitochondrion, 2017, 37, 46-54.	3.4	26
157	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
158	A lethal neonatal phenotype of mitochondrial shortâ€chain enoylâ€<scp>CoA</scp> hydrataseâ€1 deficiency. Clinical Genetics, 2017, 91, 629-633.	2.0	26
159	Gitelman-Like Syndrome Caused by Pathogenic Variants in mtDNA. Journal of the American Society of Nephrology: JASN, 2022, 33, 305-325.	6.1	26
160	Muscle 3243Aâ†'G mutation load and capacity of the mitochondrial energyâ€generating system. Annals of Neurology, 2008, 63, 473-481.	5.3	25
161	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
162	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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163	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
164	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
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166	Inhibitory effects of the beta2-adrenergic receptor agonist zilpaterol on the LPS-induced production of TNF-alpha in vitro and in vivo. Journal of Veterinary Pharmacology and Therapeutics, 2005, 28, 531-537.	1.3	24
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