Holger Prokisch

List of Publications by Year
in descending order

251 papers	26,856 citations	10956 71 h-index	7718 150 g-index
274 all docs	274 docs citations	274 times ranked	46696 citing authors

#	Article	IF	Citations
1	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). Autophagy, 2016, 12, 1-222.	4.3	4,701
2	ACSL4 dictates ferroptosis sensitivity by shaping cellular lipid composition. Nature Chemical Biology, 2017, 13, 91-98.	3.9	2,069
3	Systematic identification of trans eQTLs as putative drivers of known disease associations. Nature Genetics, 2013, 45, 1238-1243.	9.4	1,544
4	Epigenome-wide association study of body mass index, and the adverse outcomes of adiposity. Nature, 2017, 541, 81-86.	13.7	743
5	Large-scale cis- and trans-eQTL analyses identify thousands of genetic loci and polygenic scores that regulate blood gene expression. Nature Genetics, 2021, 53, 1300-1310.	9.4	590
6	The transcriptional landscape of age in human peripheral blood. Nature Communications, 2015, 6, 8570.	5.8	533
7	Systematic screen for human disease genes in yeast. Nature Genetics, 2002, 31, 400-404.	9.4	503
8	A genome-wide meta-analysis identifies 22 loci associated with eight hematological parameters in the HaemGen consortium. Nature Genetics, 2009, 41, 1182-1190.	9.4	481
9	Genetic diagnosis of Mendelian disorders via RNA sequencing. Nature Communications, 2017, 8, 15824.	5.8	432
10	Epigenome-wide association of DNA methylation markers in peripheral blood from Indian Asians and Europeans with incident type 2 diabetes: a nested case-control study. Lancet Diabetes and Endocrinology,the, 2015, 3, 526-534.	5.5	396
11	SLC2A9 influences uric acid concentrations with pronounced sex-specific effects. Nature Genetics, 2008, 40, 430-436.	9.4	363
12	COQ6 mutations in human patients produce nephrotic syndrome with sensorineural deafness. Journal of Clinical Investigation, 2011, 121, 2013-2024.	3.9	343
13	Exome Sequencing Reveals De Novo WDR45 Mutations Causing a Phenotypically Distinct, X-Linked Dominant Form of NBIA. American Journal of Human Genetics, 2012, 91, 1144-1149.	2.6	309
14	ADCK4 mutations promote steroid-resistant nephrotic syndrome through CoQ10 biosynthesis disruption. Journal of Clinical Investigation, 2013, 123, 5179-5189.	3.9	275
15	DNA methylation signatures of chronic low-grade inflammation are associated with complex diseases. Genome Biology, 2016, 17, 255.	3.8	251
16	Mitochondrial Protein Interaction Mapping Identifies Regulators of Respiratory Chain Function. Molecular Cell, 2016, 63, 621-632.	4.5	241
17	Exome sequencing identifies ACAD9 mutations as a cause of complex I deficiency. Nature Genetics, 2010, 42, 1131-1134.	9.4	234
18	Mutations in SLC39A14 disrupt manganese homeostasis and cause childhood-onset parkinsonismâ€“dystonia. Nature Communications, 2016, 7, 11601.	5.8	233

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19	Absence of an Orphan Mitochondrial Protein, C19orf12, Causes a Distinct Clinical Subtype of Neurodegeneration with Brain Iron Accumulation. American Journal of Human Genetics, 2011, 89, 543-550.	2.6	224
20	Beta-propeller protein-associated neurodegeneration: a new X-linked dominant disorder with brain iron accumulation. Brain, 2013, 136, 1708-1717.	3.7	203
21	Loss-of-function mutations in MGME1 impair mtDNA replication and cause multisystemic mitochondrial disease. Nature Genetics, 2013, 45, 214-219.	9.4	198
22	Lack of the Mitochondrial Protein Acylglycerol Kinase Causes Sengers Syndrome. American Journal of Human Genetics, 2012, 90, 314-320.	2.6	192
23	Leukoencephalopathy with thalamus and brainstem involvement and high lactate â€˜LTBLâ€™ caused by EARS2 mutations. Brain, 2012, 135, 1387-1394.	3.7	187
24	Molecular diagnosis in mitochondrial complex I deficiency using exome sequencing. Journal of Medical Genetics, 2012, 49, 277-283.	1.5	182
25	Integrative Analysis of the Mitochondrial Proteome in Yeast. PLoS Biology, 2004, 2, e160.	2.6	181
26	Deficient methylation and formylation of mt-tRNAMet wobble cytosine in a patient carrying mutations in NSUN3. Nature Communications, 2016, 7, 12039.	5.8	178
27	Exome Sequence Reveals Mutations in CoA Synthase as a Cause of Neurodegeneration with Brain Iron Accumulation. American Journal of Human Genetics, 2014, 94, 11-22.	2.6	176
28	Characterization of whole-genome autosomal differences of DNA methylation between men and women. Epigenetics and Chromatin, 2015, 8, 43.	1.8	176
29	Novel (ovario) leukodystrophy related to AARS2 mutations. Neurology, 2014, 82, 2063-2071.	1.5	172
30	Mutations of the Mitochondrial-tRNA Modifier MTO1 Cause Hypertrophic Cardiomyopathy and Lactic Acidosis. American Journal of Human Genetics, 2012, 90, 1079-1087.	2.6	164
31	Mitochondrial membrane protein associated neurodegenration: A novel variant of neurodegeneration with brain iron accumulation. Movement Disorders, 2013, 28, 224-227.	2.2	162
32	DNA Methylation of Lipid-Related Genes Affects Blood Lipid Levels. Circulation: Cardiovascular Genetics, 2015, 8, 334-342.	5.1	151
33	Identification of Novel Genetic Loci Associated with Thyroid Peroxidase Antibodies and Clinical Thyroid Disease. PLoS Genetics, 2014, 10, e1004123.	1.5	150
34	Genetics of mitochondrial diseases: Identifying mutations to help diagnosis. EBioMedicine, 2020, 56, 102784.	2.7	145
35	Monogenic variants in dystonia: an exome-wide sequencing study. Lancet Neurology, The, 2020, 19, 908-918.	4.9	139
36	Mutations in FBXL4, Encoding a Mitochondrial Protein, Cause Early-Onset Mitochondrial Encephalomyopathy. American Journal of Human Genetics, 2013, 93, 482-495.	2.6	138

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37	Expression analysis of dopaminergic neurons in Parkinsonâ€™s disease and aging links transcriptional dysregulation of energy metabolism to cell death. Acta Neuropathologica, 2011, 122, 75-86.	3.9	127
38	ELAC2 Mutations Cause a Mitochondrial RNA Processing Defect Associated with Hypertrophic Cardiomyopathy. American Journal of Human Genetics, 2013, 93, 211-223.	2.6	127
39	Biallelic Mutations in DNAJC12 Cause Hyperphenylalaninemia, Dystonia, and Intellectual Disability. American Journal of Human Genetics, 2017, 100, 257-266.	2.6	127
40	Mutations in GTPBP3 Cause a Mitochondrial Translation Defect Associated with Hypertrophic Cardiomyopathy, Lactic Acidosis, and Encephalopathy. American Journal of Human Genetics, 2014, 95, 708-720.	2.6	123
41	Riboflavin-Responsive and -Non-responsive Mutations in FAD Synthase Cause Multiple Acyl-CoA Dehydrogenase and Combined Respiratory-Chain Deficiency. American Journal of Human Genetics, 2016, 98, 1130-1145.	2.6	118
42	Cell Specific eQTL Analysis without Sorting Cells. PLoS Genetics, 2015, 11, e1005223.	1.5	115
43	Riboflavin-responsive oxidative phosphorylation complex I deficiency caused by defective ACAD9: new function for an old gene. Brain, 2011, 134, 210-219.	3.7	113
44	OUTRIDER: A Statistical Method for Detecting Aberrantly Expressed Genes in RNA Sequencing Data. American Journal of Human Genetics, 2018, 103, 907-917.	2.6	112
45	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.	2.6	111
46	Biallelic Mutations in NBAS Cause Recurrent Acute Liver Failure with Onset in Infancy. American Journal of Human Genetics, 2015, 97, 163-169.	2.6	110
47	Thiamine Pyrophosphokinase Deficiency in Encephalopathic Children with Defects in the Pyruvate Oxidation Pathway. American Journal of Human Genetics, 2011, 89, 806-812.	2.6	107
48	A Meta-analysis of Gene Expression Signatures of Blood Pressure and Hypertension. PLoS Genetics, 2015, 11, e1005035.	1.5	107
49	CADmutations and uridine-responsive epileptic encephalopathy. Brain, 2017, 140, 279-286.	3.7	106
50	Disturbed mitochondrial and peroxisomal dynamics due to loss of MFF causes Leigh-like encephalopathy, optic atrophy and peripheral neuropathy. Journal of Medical Genetics, 2016, 53, 270-278.	1.5	105
51	A Genome-wide Association Study Identifies Three Loci Associated with Mean Platelet Volume. American Journal of Human Genetics, 2009, 84, 66-71.	2.6	104
52	Recessive mutations in VPS13D cause childhood onset movement disorders. Annals of Neurology, 2018, 83, 1089-1095.	2.8	104
53	An Integrated Epigenetic and Transcriptomic Analysis Reveals Distinct Tissue-Specific Patterns of DNA Methylation Associated with Atopic Dermatitis. Journal of Investigative Dermatology, 2014, 134, 1873-1883.	0.3	103
54	Spectrum of combined respiratory chain defects. Journal of Inherited Metabolic Disease, 2015, 38, 629-640.	1.7	102

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55	Absence of BiP Co-chaperone DNAJC3 Causes Diabetes Mellitus and Multisystemic Neurodegeneration. American Journal of Human Genetics, 2014, 95, 689-697.	2.6	100
56	Absence of the Autophagy Adaptor SQSTM1/p62 Causes Childhood-Onset Neurodegeneration with Ataxia, Dystonia, and Gaze Palsy. American Journal of Human Genetics, 2016, 99, 735-743.	2.6	99
57	The Human Blood Metabolome-Transcriptome Interface. PLoS Genetics, 2015, 11, e1005274.	1.5	99
58	DHTKD1 Mutations Cause 2-Aminoadipic and 2-Oxoadipic Aciduria. American Journal of Human Genetics, 2012, 91, 1082-1087.	2.6	94
59	Recurrent acute liver failure due to NBAS deficiency: phenotypic spectrum, disease mechanisms, and therapeutic concepts. Journal of Inherited Metabolic Disease, 2016, 39, 3-16.	1.7	92
60	Deficiency of <scp>ECHS</scp>1 causes mitochondrial encephalopathy with cardiac involvement. Annals of Clinical and Translational Neurology, 2015, 2, 492-509.	1.7	90
61	Impaired complex I repair causes recessive Leberâ€™s hereditary optic neuropathy. Journal of Clinical Investigation, 2021, 131, .	3.9	89
62	Biallelic Variants in UBA5 Reveal that Disruption of the UFM1 Cascade Can Result in Early-Onset Encephalopathy. American Journal of Human Genetics, 2016, 99, 695-703.	2.6	87
63	VARS2andTARS2Mutations in Patients with Mitochondrial Encephalomyopathies. Human Mutation, 2014, 35, 983-989.	1.1	86
64	COQ4 Mutations Cause a Broad Spectrum of Mitochondrial Disorders Associated with CoQ10 Deficiency. American Journal of Human Genetics, 2015, 96, 309-317.	2.6	86
65	Human thioredoxin 2 deficiency impairs mitochondrial redox homeostasis and causes early-onset neurodegeneration. Brain, 2016, 139, 346-354.	3.7	86
66	GWAS on longitudinal growth traits reveals different genetic factors influencing infant, child, and adult BMI. Science Advances, 2019, 5, eaaw3095.	4.7	86
67	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38.	3.6	85
68	Homozygous missense mutation in BOLA3 causes multiple mitochondrial dysfunctions syndrome in two siblings. Journal of Inherited Metabolic Disease, 2013, 36, 55-62.	1.7	83
69	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.	2.6	83
70	Clinical, biochemical, and genetic spectrum of seven patients with NFU1 deficiency. Frontiers in Genetics, 2015, 06, 123.	1.1	81
71	LRPPRCmutations cause early-onset multisystem mitochondrial disease outside of the French-Canadian population. Brain, 2015, 138, 3503-3519.	3.7	81
72	Association between DNA Methylation in Whole Blood and Measures of Glucose Metabolism: KORA F4 Study. PLoS ONE, 2016, 11, e0152314.	1.1	81

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73	Mutation screening of 75 candidate genes in 152 complex I deficiency cases identifies pathogenic variants in 16 genes includingNDUFB9. Journal of Medical Genetics, 2012, 49, 83-89.	1.5	78
74	Detection of aberrant splicing events in RNA-seq data using FRASER. Nature Communications, 2021, 12, 529.	5.8	78
75	Bi-allelic Truncating Mutations in TANGO2 Cause Infancy-Onset Recurrent Metabolic Crises with Encephalocardiomyopathy. American Journal of Human Genetics, 2016, 98, 358-362.	2.6	77
76	Mitochondria: Much ado about nothing? How dangerous is reactive oxygen species production?. International Journal of Biochemistry and Cell Biology, 2015, 63, 16-20.	1.2	76
77	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.	0.5	76
78	A Whole-Blood Transcriptome Meta-Analysis Identifies Gene Expression Signatures of Cigarette Smoking. Human Molecular Genetics, 2016, 25, ddw288.	1.4	76
79	Mutations inSDHDlead to autosomal recessive encephalomyopathy and isolated mitochondrial complex II deficiency. Journal of Medical Genetics, 2014, 51, 170-175.	1.5	75
80	NAXE Mutations Disrupt the Cellular NAD(P)HX Repair System and Cause a Lethal Neurometabolic Disorder of Early Childhood. American Journal of Human Genetics, 2016, 99, 894-902.	2.6	75
81	Biallelic IARS Mutations Cause Growth Retardation with Prenatal Onset, Intellectual Disability, Muscular Hypotonia, and Infantile Hepatopathy. American Journal of Human Genetics, 2016, 99, 414-422.	2.6	73
82	Delineating MT-ATP6 -associated disease. Neurology: Genetics, 2020, 6, e393.	0.9	73
83	Impaired riboflavin transport due to missense mutations in SLC52A2 causes Brownâ€Vialettoâ€Van Laere syndrome. Journal of Inherited Metabolic Disease, 2012, 35, 943-948.	1.7	72
84	Analyzing Illumina Gene Expression Microarray Data from Different Tissues: Methodological Aspects of Data Analysis in the MetaXpress Consortium. PLoS ONE, 2012, 7, e50938.	1.1	71
85	MitoP2: An Integrative Tool for the Analysis of the Mitochondrial Proteome. Molecular Biotechnology, 2008, 40, 306-315.	1.3	69
86	Assessing Systems Properties of Yeast Mitochondria through an Interaction Map of the Organelle. PLoS Genetics, 2006, 2, e170.	1.5	67
87	MTO1 Mutations are Associated with Hypertrophic Cardiomyopathy and Lactic Acidosis and Cause Respiratory Chain Deficiency in Humans and Yeast. Human Mutation, 2013, 34, 1501-1509.	1.1	67
88	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.	0.5	65
89	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. Journal of Clinical Investigation, 2019, 130, 108-125.	3.9	65
90	Mutations in APOPT1, Encoding a Mitochondrial Protein, Cause Cavitating Leukoencephalopathy with Cytochrome c Oxidase Deficiency. American Journal of Human Genetics, 2014, 95, 315-325.	2.6	64

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91	Assessing Mitochondrial Bioenergetics in Isolated Mitochondria from Various Mouse Tissues Using Seahorse XF96 Analyzer. Methods in Molecular Biology, 2017, 1567, 217-230.	0.4	64
92	Mutations in MDH2, Encoding a Krebs Cycle Enzyme, Cause Early-Onset Severe Encephalopathy. American Journal of Human Genetics, 2017, 100, 151-159.	2.6	63
93	Quantification and discovery of sequence determinants of proteinâ€perâ€mRNA amount inÂ29Âhuman tissues. Molecular Systems Biology, 2019, 15, e8513.	3.2	63
94	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. Orphanet Journal of Rare Diseases, 2018, 13, 120.	1.2	61
95	A Guideline for the Diagnosis of Pediatric Mitochondrial Disease: The Value of Muscle and Skin Biopsies in the Genetics Era. Neuropediatrics, 2017, 48, 309-314.	0.3	60
96	Genetic variation influencing DNA methylation provides insights into molecular mechanisms regulating genomic function. Nature Genetics, 2022, 54, 18-29.	9.4	60
97	CoQ deficiency causes disruption of mitochondrial sulfide oxidation, a new pathomechanism associated with this syndrome. EMBO Molecular Medicine, 2017, 9, 78-95.	3.3	59
98	Macrocytic Anemia and Mitochondriopathy Resulting from a Defect in Sideroflexin 4. American Journal of Human Genetics, 2013, 93, 906-914.	2.6	58
99	Biallelic C1QBP Mutations Cause Severe Neonatal-, Childhood-, or Later-Onset Cardiomyopathy Associated with Combined Respiratory-Chain Deficiencies. American Journal of Human Genetics, 2017, 101, 525-538.	2.6	58
100	ncRNAs: New Players in Mitochondrial Health and Disease?. Frontiers in Genetics, 2020, 11, 95.	1.1	58
101	Detection of aberrant gene expression events in RNA sequencing data. Nature Protocols, 2021, 16, 1276-1296.	5.5	58
102	Mutations of C19orf12, coding for a transmembrane glycine zipper containing mitochondrial protein, cause mis-localization of the protein, inability to respond to oxidative stress and increased mitochondrial Ca2+. Frontiers in Genetics, 2015, 6, 185.	1.1	57
103	Biallelic Mutations in TMEM126B Cause Severe Complex I Deficiency with a Variable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 217-227.	2.6	57
104	Treatable mitochondrial diseases: cofactor metabolism and beyond. Brain, 2017, 140, e11-e11.	3.7	57
105	Mitochondrial DNA mutation analysis from exome sequencingâ€”A more holistic approach in diagnostics of suspected mitochondrial disease. Journal of Inherited Metabolic Disease, 2019, 42, 909-917.	1.7	57
106	Biallelic Mutations in LIPT2 Cause a Mitochondrial Lipoylation Defect Associated with Severe Neonatal Encephalopathy. American Journal of Human Genetics, 2017, 101, 283-290.	2.6	55
107	OCR-Stats: Robust estimation and statistical testing of mitochondrial respiration activities using Seahorse XF Analyzer. PLoS ONE, 2018, 13, e0199938.	1.1	55
108	Complex I assembly function and fatty acid oxidation enzyme activity of ACAD9 both contribute to disease severity in ACAD9 deficiency. Human Molecular Genetics, 2015, 24, 3238-3247.	1.4	53

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109	The Dimensions of Primary Mitochondrial Disorders. Frontiers in Cell and Developmental Biology, 2020, 8, 600079.	1.8	53
110	Advancing genomic approaches to the molecular diagnosis of mitochondrial disease. Essays in Biochemistry, 2018, 62, 399-408.	2.1	51
111	NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. Brain, 2019, 142, 50-58.	3.7	51
112	Expanding the clinical and molecular spectrum of thiamine pyrophosphokinase deficiency: A treatable neurological disorder caused by TPK1 mutations. Molecular Genetics and Metabolism, 2014, 113, 301-306.	0.5	50
113	SCYL1 variants cause a syndrome with lowÎ³-glutamyl-transferase cholestasis, acute liver failure, and neurodegeneration (CALFAN). Genetics in Medicine, 2018, 20, 1255-1265.	1.1	50
114	Singleâ€cell expression profiling of dopaminergic neurons combined with association analysis identifies pyridoxal kinase as Parkinson's disease gene. Annals of Neurology, 2009, 66, 792-798.	2.8	49
115	Sudden Cardiac Death Due to Deficiency of the Mitochondrial Inorganic Pyrophosphatase PPA2. American Journal of Human Genetics, 2016, 99, 674-682.	2.6	48
116	The diagnosis of inborn errors of metabolism by an integrative â€œmultiâ€omicsâ€•approach: A perspective encompassing genomics, transcriptomics, and proteomics. Journal of Inherited Metabolic Disease, 2020, 43, 25-35.	1.7	47
117	Defining clinical subgroups and genotypeâ€“phenotype correlations in NBAS-associated disease across 110 patients. Genetics in Medicine, 2020, 22, 610-621.	1.1	46
118	Clinical, morphological, biochemical, imaging and outcome parameters in 21 individuals with mitochondrial maintenance defect related to FBXL4 mutations. Journal of Inherited Metabolic Disease, 2015, 38, 905-914.	1.7	45
119	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.	1.4	45
120	The spectrum of pyruvate oxidation defects in the diagnosis of mitochondrial disorders. Journal of Inherited Metabolic Disease, 2015, 38, 391-403.	1.7	44
121	MRPL44 mutations cause a slowly progressive multisystem disease with childhood-onset hypertrophic cardiomyopathy. Neurogenetics, 2015, 16, 319-323.	0.7	44
122	Impact of common regulatory single-nucleotide variants on gene expression profiles in whole blood. European Journal of Human Genetics, 2013, 21, 48-54.	1.4	43
123	MTO1 mediates tissue specificity of OXPHOS defects via tRNA modification and translation optimization, which can be bypassed by dietary intervention. Human Molecular Genetics, 2015, 24, 2247-2266.	1.4	43
124	Mutations in PPCS, Encoding Phosphopantothenoylcysteine Synthetase, Cause Autosomal-Recessive Dilated Cardiomyopathy. American Journal of Human Genetics, 2018, 102, 1018-1030.	2.6	42
125	BPAN. International Review of Neurobiology, 2013, 110, 85-90.	0.9	41
126	Clinical Features, Molecular Heterogeneity, and Prognostic Implications in YARS2-Related Mitochondrial Myopathy. JAMA Neurology, 2017, 74, 686.	4.5	41

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127	Molecular and clinical spectra of FBXL4 deficiency. Human Mutation, 2017, 38, 1649-1659.	1.1	41
128	Bi-allelic Mutations in NDUFA6 Establish Its Role in Early-Onset Isolated Mitochondrial Complex I Deficiency. American Journal of Human Genetics, 2018, 103, 592-601.	2.6	41
129	Genetic Determinants of Circulating Interleukin-1 Receptor Antagonist Levels and Their Association With Glycemic Traits. Diabetes, 2014, 63, 4343-4359.	0.3	40
130	Extensive alterations of the whole-blood transcriptome are associated with body mass index: results of an mRNA profiling study involving two large population-based cohorts. BMC Medical Genomics, 2015, 8, 65.	0.7	40
131	NDUFB8 Mutations Cause Mitochondrial Complex I Deficiency in Individuals with Leigh-like Encephalomyopathy. American Journal of Human Genetics, 2018, 102, 460-467.	2.6	40
132	Bi-allelic ADPRHL2 Mutations Cause Neurodegeneration with Developmental Delay, Ataxia, and Axonal Neuropathy. American Journal of Human Genetics, 2018, 103, 817-825.	2.6	40
133	Elevated glutaric acid levels in Dhtkd1-/Gcdh- double knockout mice challenge our current understanding of lysine metabolism. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2017, 1863, 2220-2228.	1.8	39
134	Bi-allelic mutations in TRAPPC2L result in a neurodevelopmental disorder and have an impact on RAB11 in fibroblasts. Journal of Medical Genetics, 2018, 55, 753-764.	1.5	39
135	RINT1 Bi-allelic Variations Cause Infantile-Onset Recurrent Acute Liver Failure and Skeletal Abnormalities. American Journal of Human Genetics, 2019, 105, 108-121.	2.6	39
136	Pathogenic Bi-allelic Mutations in NDUFAF8 Cause Leigh Syndrome with an Isolated Complex I Deficiency. American Journal of Human Genetics, 2020, 106, 92-101.	2.6	39
137	â€œTranscriptomicsâ€• molecular diagnosis of inborn errors of metabolism via RNAâ€•sequencing. Journal of Inherited Metabolic Disease, 2018, 41, 525-532.	1.7	38
138	A Population-Based Epidemiological and Genetic Study of X-Linked Retinitis Pigmentosa. , 2007, 48, 4012.		36
139	Infantile Leigh-like syndrome caused by SLC19A3 mutations is a treatable disease. Brain, 2014, 137, e295-e295.	3.7	36
140	Bi-Allelic UQCRFS1 Variants Are Associated with Mitochondrial Complex III Deficiency, Cardiomyopathy, and Alopecia Totalis. American Journal of Human Genetics, 2020, 106, 102-111.	2.6	36
141	DNA methylation and lipid metabolism: an EWAS of 226 metabolic measures. Clinical Epigenetics, 2021, 13, 7.	1.8	36
142	Genetic basis of mitochondrial diseases. FEBS Letters, 2021, 595, 1132-1158.	1.3	36
143	Bezafibrate Improves Insulin Sensitivity and Metabolic Flexibility in STZ-Induced Diabetic Mice. Diabetes, 2016, 65, 2540-2552.	0.3	35
144	Bainbridgeâ€“Ropers syndrome caused by loss-of-function variants in ASXL3: a recognizable condition. European Journal of Human Genetics, 2017, 25, 183-191.	1.4	35

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145	Lifetime risk of autosomal recessive mitochondrial disorders calculated from genetic databases. EBioMedicine, 2020, 54, 102730.	2.7	35
146	A mutation screening of oncogenes, tumor suppressor gene TP53 and nuclear encoded mitochondrial complex I genes in oncocytic thyroid tumors. BMC Cancer, 2015, 15, 157.	1.1	34
147	MRPS22 mutation causes fatal neonatal lactic acidosis with brain and heart abnormalities. Neurogenetics, 2015, 16, 237-240.	0.7	34
148	Transcriptome-Wide Analysis Identifies Novel Associations With Blood Pressure. Hypertension, 2017, 70, 743-750.	1.3	34
149	Bi-allelic Mutations in Phe-tRNA Synthetase Associated with a Multi-system Pulmonary Disease Support Non-translational Function. American Journal of Human Genetics, 2018, 103, 100-114.	2.6	34
150	Combined Respiratory Chain Deficiency and UQCC2 Mutations in Neonatal Encephalomyopathy: Defective Supercomplex Assembly in Complex III Deficiencies. Oxidative Medicine and Cellular Longevity, 2017, 2017, 1-11.	1.9	33
151	Association of Methylation Signals With Incident Coronary Heart Disease in an Epigenome-Wide Assessment of Circulating Tumor Necrosis Factor Î±. JAMA Cardiology, 2018, 3, 463.	3.0	33
152	The genetics of mitochondrial disease: dissecting mitochondrial pathology using multiâ€omic pipelines. Journal of Pathology, 2021, 254, 430-442.	2.1	33
153	A truncating PET100 variant causing fatal infantile lactic acidosis and isolated cytochrome c oxidase deficiency. European Journal of Human Genetics, 2015, 23, 935-939.	1.4	32
154	Mapping the Genetic Architecture of Gene Regulation in Whole Blood. PLoS ONE, 2014, 9, e93844.	1.1	31
155	A recurrent mitochondrial p.Trp22ArgNDUFB3variant causes a distinctive facial appearance, short stature and a mild biochemical and clinical phenotype. Journal of Medical Genetics, 2016, 53, 634-641.	1.5	31
156	Mutations inELAC2associated with hypertrophic cardiomyopathy impair mitochondrial tRNA 3â€²â€end processing. Human Mutation, 2019, 40, 1731-1748.	1.1	31
157	Meta-analyses identify DNA methylation associated with kidney function and damage. Nature Communications, 2021, 12, 7174.	5.8	30
158	Mutation in ITCH Gene Can Cause Syndromic Multisystem Autoimmune Disease With Acute Liver Failure. Pediatrics, 2019, 143, .	1.0	29
159	Mitochondrial Regulation of the 26S Proteasome. Cell Reports, 2020, 32, 108059.	2.9	28
160	Impairment of Drosophila Orthologs of the Human Orphan Protein C19orf12 Induces Bang Sensitivity and Neurodegeneration. PLoS ONE, 2014, 9, e89439.	1.1	28
161	High incidence and variable clinical outcome of cardiac hypertrophy due to ACAD9 mutations in childhood. European Journal of Human Genetics, 2016, 24, 1112-1116.	1.4	27
162	Neonatal encephalocardiomyopathy caused by mutations in VARS2. Metabolic Brain Disease, 2017, 32, 267-270.	1.4	26

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163	Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults. JAMA Neurology, 2018, 75, 105.	4.5	26
164	Mutations in TTC19: expanding the molecular, clinical and biochemical phenotype. Orphanet Journal of Rare Diseases, 2015, 10, 40.	1.2	25
165	Severe respiratory complex III defect prevents liver adaptation to prolonged fasting. Journal of Hepatology, 2016, 65, 377-385.	1.8	25
166	Identification of Disease-Causing Mutations by Functional Complementation of Patient-Derived Fibroblast Cell Lines. Methods in Molecular Biology, 2017, 1567, 391-406.	0.4	25
167	Detection of 6-demethoxyubiquinone in CoQ10 deficiency disorders: Insights into enzyme interactions and identification of potential therapeutics. Molecular Genetics and Metabolism, 2017, 121, 216-223.	0.5	25
168	Biallelic variants in the transcription factor PAX7 are a new genetic cause of myopathy. Genetics in Medicine, 2019, 21, 2521-2531.	1.1	25
169	Biallelic variants inWARS2encoding mitochondrial tryptophanyl-tRNA synthase in six individuals with mitochondrial encephalopathy. Human Mutation, 2017, 38, 1786-1795.	1.1	24
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