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	Whole genome and exome sequencing identify NDUFV2 mutations as a new cause of progressive cavitating leukoencephalopathy. Journal of Medical Genetics, 2022, 59, 351-357.	1.5	5
2	Genetic variation influencing DNA methylation provides insights into molecular mechanisms regulating genomic function. Nature Genetics, 2022, 54, 18-29.	9.4	60
3	Population-based screening in children for early diagnosis and treatment of familial hypercholesterolemia: design of the VRONI study. European Journal of Public Health, 2022, 32, 422-428.	0.1	11
4	Variants in Mitochondrial <scp>ATP</scp> Synthase Cause Variable Neurologic Phenotypes. Annals of Neurology, 2022, 91, 225-237.	2.8	12
5	Leigh Syndrome: A Study of 209 Patients at the Beijing Children's Hospital. Annals of Neurology, 2022, 91, 466-482.	2.8	10
6	Identification and characterization of novel <scp>MPC1</scp> gene variants causing mitochondrial pyruvate carrier deficiency. Journal of Inherited Metabolic Disease, 2022, 45, 264-277.	1.7	7
7	Clinicoâ€radiological features, molecular spectrum, and identification of prognostic factors in developmental and epileptic encephalopathy due to inosine triphosphate pyrophosphatase (ITPase) deficiency. Human Mutation, 2022, 43, 403-419.	1.1	9
8	DNAJC30 defect: a frequent cause of recessive Leber hereditary optic neuropathy and Leigh syndrome. Brain, 2022, 145, 1624-1631.	3.7	21
9	RNA sequencing role and application in clinical diagnostic. Pediatric Investigation, 2022, 6, 29-35.	0.6	12
10	Protonation-Dependent Sequencing of 5-Formylcytidine in RNA. Biochemistry, 2022, 61, 535-544.	1.2	10
11	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38.	3.6	85
12	AOPEP variants as a novel cause of recessive dystonia: Generalized dystonia and dystonia-parkinsonism. Parkinsonism and Related Disorders, 2022, 97, 52-56.	1.1	7
13	Population-based screening in children for early diagnosis and treatment of familial hypercholesterolemia: design of the VRONI study. Medizinische Genetik, 2022, 34, 41-51.	0.1	0
14	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.	1.7	20
15	Guidelines for clinical interpretation of variant pathogenicity using RNA phenotypes. Human Mutation, 2022, 43, 1056-1070.	1.1	8
16	Detection of aberrant splicing events in RNA-seq data using FRASER. Nature Communications, 2021, 12, 529.	5.8	78
17	Pediatric <scp>Leigh</scp> Syndrome: Neuroimaging Features and Genetic Correlations. Annals of Neurology, 2021, 89, 629-631.	2.8	4
18	X-Linked Retinitis Pigmentosa Caused by Non-Canonical Splice Site Variants in RPGR. International Journal of Molecular Sciences, 2021, 22, 850.	1.8	6

#	Article	IF	Citations
19	Detection of aberrant gene expression events in RNA sequencing data. Nature Protocols, 2021, 16, 1276-1296.	5.5	58
20	DNA methylation and lipid metabolism: an EWAS of 226 metabolic measures. Clinical Epigenetics, 2021, 13, 7.	1.8	36
21	Expanding the clinical and genetic spectrum of FDXR deficiency by functional validation of variants of uncertain significance. Human Mutation, 2021, 42, 310-319.	1.1	11
22	Epigenome-wide association study of whole blood gene expression in Framingham Heart Study participants provides molecular insight into the potential role of CHRNA5 in cigarette smoking-related lung diseases. Clinical Epigenetics, 2021, 13, 60.	1.8	14
23	The genetics of mitochondrial disease: dissecting mitochondrial pathology using multiâ€omic pipelines. Journal of Pathology, 2021, 254, 430-442.	2.1	33
24	Novel NDUFA12 variants are associated with isolated complex I defect and variable clinical manifestation. Human Mutation, 2021, 42, 699-710.	1.1	12
25	Genetic basis of mitochondrial diseases. FEBS Letters, 2021, 595, 1132-1158.	1.3	36
26	Impaired complex I repair causes recessive Leberâ€™s hereditary optic neuropathy. Journal of Clinical Investigation, 2021, 131, .	3.9	89
27	Identification of a Novel Variant in MT-CO3 Causing MELAS. Frontiers in Genetics, 2021, 12, 638749.	1.1	2
28	A comprehensive phenotypic characterization of a whole-body Wdr45 knock-out mouse. Mammalian Genome, 2021, 32, 332-349.	1.0	4
29	Aberrant activity of mitochondrial NCLX is linked to impaired synaptic transmission and is associated with mental retardation. Communications Biology, 2021, 4, 666.	2.0	22
30	Comparison of genetic risk prediction models to improve prediction of coronary heart disease in two large cohorts of the MONICA/KORA study. Genetic Epidemiology, 2021, 45, 633-650.	0.6	6
31	Myopathic mitochondrial DNA depletion syndrome associated with biallelic variants in LIG3. Brain, 2021, 144, e74-e74.	3.7	5
32	How Machine Learning and Statistical Models Advance Molecular Diagnostics of Rare Disorders Via Analysis of RNA Sequencing Data. Frontiers in Molecular Biosciences, 2021, 8, 647277.	1.6	12
33	Biallelic COA7-Variants Leading to Developmental Regression With Progressive Spasticity and Brain Atrophy in a Chinese Patient. Frontiers in Genetics, 2021, 12, 685035.	1.1	7
34	Muscular and Molecular Pathology Associated with SPATA5 Deficiency in a Child with EHLMRS. International Journal of Molecular Sciences, 2021, 22, 7835.	1.8	4
35	NBAS Variants Are Associated with Quantitative and Qualitative NK and B Cell Deficiency. Journal of Clinical Immunology, 2021, 41, 1781-1793.	2.0	10
36	Pathogenic variants in MRPL44 cause infantile cardiomyopathy due to a mitochondrial translation defect. Molecular Genetics and Metabolism, 2021, 133, 362-371.	0.5	5

#	Article	IF	Citations
37	Prevalence and clinical prediction of mitochondrial disorders in a large neuropediatric cohort. Clinical Genetics, 2021, 100, 766-770.	1.0	5
38	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
39	A novel cryptic splice site mutation in COL1A2 as a cause of osteogenesis imperfecta. Bone Reports, 2021, 15, 101110.	0.2	3
40	Identification of a Novel m.3955G>A Variant in MT-ND1 Associated with Leigh Syndrome. Mitochondrion, 2021, 62, 13-23.	1.6	3
41	Multi-Omics Approach to Mitochondrial DNA Damage in Human Muscle Fibers. International Journal of Molecular Sciences, 2021, 22, 11080.	1.8	2
42	Characterising a homozygous twoâ€exon deletion in UQCRH : comparing human and mouse phenotypes. EMBO Molecular Medicine, 2021, 13, e14397.	3.3	5
43	Mitochondrial disorders. Deutsches Ärzteblatt International, 2021, , .	0.6	12
44	ATP synthase deficiency due to m.8528T>C mutationÂâ€“ a novel cause of severe neonatal hyperammonemia requiring hemodialysis. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 389-393.	0.4	0
45	Inactivity of Peptidase ClpP Causes Primary Accumulation of Mitochondrial Disaggregase ClpX with Its Interacting Nucleoid Proteins, and of mtDNA. Cells, 2021, 10, 3354.	1.8	4
46	Mitochondrial Transporter Defects: Successful Treatment with Ketogenic Diet Therapy. Neuropediatrics, 2021, 52, .	0.3	0
47	Epigenome-wide association study of serum urate reveals insights into urate co-regulation and the SLC2A9 locus. Nature Communications, 2021, 12, 7173.	5.8	8
48	Meta-analyses identify DNA methylation associated with kidney function and damage. Nature Communications, 2021, 12, 7174.	5.8	30
49	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
50	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
51	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
52	Defining clinical subgroups and genotypeâ€“phenotype correlations in NBAS-associated disease across 110 patients. Genetics in Medicine, 2020, 22, 610-621.	1.1	46
53	NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. Brain, 2020, 143, e8-e8.	3.7	18
54	Coenzyme Q10 modulates sulfide metabolism and links the mitochondrial respiratory chain to pathways associated to one carbon metabolism. Human Molecular Genetics, 2020, 29, 3296-3311.	1.4	16

#	Article	IF	Citations
55	Monogenic variants in dystonia: an exome-wide sequencing study. Lancet Neurology, The, 2020, 19, 908-918.	4.9	139
56	Case Report: Rapid Treatment of Uridine-Responsive Epileptic Encephalopathy Caused by CAD Deficiency. Frontiers in Pharmacology, 2020, 11, 608737.	1.6	8
57	PRPS1 loss-of-function variants, from isolated hearing loss to severe congenital encephalopathy: New cases and literature review. European Journal of Medical Genetics, 2020, 63, 104033.	0.7	11
58	PKAN neurodegeneration and residual PANK2 activities in patient erythrocytes. Annals of Clinical and Translational Neurology, 2020, 7, 1340-1351.	1.7	11
59	Genotypic diversity and phenotypic spectrum of infantile liver failure syndrome type 1 due to variants in LARS1. Genetics in Medicine, 2020, 22, 1863-1873.	1.1	19
60	Paroxysmal and non-paroxysmal dystonia in 3 patients with biallelic ECHS1 variants: Expanding the neurological spectrum and therapeutic approaches. European Journal of Medical Genetics, 2020, 63, 104046.	0.7	12
61	Mitochondrial Regulation of the 26S Proteasome. Cell Reports, 2020, 32, 108059.	2.9	28
62	Rescue of respiratory failure in pulmonary alveolar proteinosis due to pathogenic MARS1 variants. Pediatric Pulmonology, 2020, 55, 3057-3066.	1.0	19
63	The Dimensions of Primary Mitochondrial Disorders. Frontiers in Cell and Developmental Biology, 2020, 8, 600079.	1.8	53
64	Recurrent acute liver failure in alanyl-tRNA synthetase-1 (AARS1) deficiency. Molecular Genetics and Metabolism Reports, 2020, 25, 100681.	0.4	2
65	Genetics of mitochondrial diseases: Identifying mutations to help diagnosis. EBioMedicine, 2020, 56, 102784.	2.7	145
66	Delineating MT-ATP6 -associated disease. Neurology: Genetics, 2020, 6, e393.	0.9	73
67	ncRNAs: New Players in Mitochondrial Health and Disease?. Frontiers in Genetics, 2020, 11, 95.	1.1	58
68	Lifetime risk of autosomal recessive mitochondrial disorders calculated from genetic databases. EBioMedicine, 2020, 54, 102730.	2.7	35
69	Blood DNA methylation sites predict death risk in a longitudinal study of 12, 300 individuals. Aging, 2020, 12, 14092-14124.	1.4	15
70	The Clinical Application of RNA Sequencing in Genetic Diagnosis of Mendelian Disorders. Clinics in Laboratory Medicine, 2020, 40, 121-133.	0.7	19
71	Molecular diagnostics of Mendelian disorders via combined DNA and RNA sequencing. Medizinische Genetik, 2019, 31, 191-197.	0.1	0
72	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

#	Article	IF	Citations
73	Arabidopsis thaliana alternative dehydrogenases: a potential therapy for mitochondrial complex I deficiency? Perspectives and pitfalls. Orphanet Journal of Rare Diseases, 2019, 14, 236.	1.2	11
74	GWAS on longitudinal growth traits reveals different genetic factors influencing infant, child, and adult BMI. Science Advances, 2019, 5, eaaw3095.	4.7	86
75	Biallelic variants in the transcription factor PAX7 are a new genetic cause of myopathy. Genetics in Medicine, 2019, 21, 2521-2531.	1.1	25
76	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
77	Mutations inELAC2associated with hypertrophic cardiomyopathy impair mitochondrial tRNA 3â€²â€end processing. Human Mutation, 2019, 40, 1731-1748.	1.1	31
78	Mitochondrial Disease Genetics. , 2019, , 41-62.		0
79	Quantification and discovery of sequence determinants of proteinâ€perâ€mRNA amount inÂ29Âhuman tissues. Molecular Systems Biology, 2019, 15, e8513.	3.2	63
80	The metabolic network coherence of human transcriptomes is associated with genetic variation at the cadherin 18 locus. Human Genetics, 2019, 138, 375-388.	1.8	6
81	Homozygous frame shift variant in ATP7B exon 1 leads to bypass of nonsense-mediated mRNA decay and to a protein capable of copper export. European Journal of Human Genetics, 2019, 27, 879-887.	1.4	6
82	Mutation in ITCH Gene Can Cause Syndromic Multisystem Autoimmune Disease With Acute Liver Failure. Pediatrics, 2019, 143, .	1.0	29
83	NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. Brain, 2019, 142, 50-58.	3.7	51
84	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. Journal of Clinical Investigation, 2019, 130, 108-125.	3.9	65
85	Genetic Basis of Mitochondrial Cardiomyopathy. Cardiac and Vascular Biology, 2019, , 93-139.	0.2	1
86	Recessive mutations in VPS13D cause childhood onset movement disorders. Annals of Neurology, 2018, 83, 1089-1095.	2.8	104
87	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
88	â€œTranscriptomicsâ€• molecular diagnosis of inborn errors of metabolism via RNAâ€sequencing. Journal of Inherited Metabolic Disease, 2018, 41, 525-532.	1.7	38
89	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
90	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

#	Article	IF	Citations
91	Clinical, biochemical, and genetic features associated with VARS2 -related mitochondrial disease. Human Mutation, 2018, 39, 563-578.	1.1	22
92	Severe ichthyosis in MPDU1â€CDG. Journal of Inherited Metabolic Disease, 2018, 41, 1293-1294.	1.7	8
93	Biallelic Mutations in SLC1A2; an Additional Mode of Inheritance for SLC1A2-Related Epilepsy. Neuropediatrics, 2018, 49, 059-062.	0.3	14
94	Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults. JAMA Neurology, 2018, 75, 105.	4.5	26
95	The genotypic and phenotypic spectrum of MTO1 deficiency. Molecular Genetics and Metabolism, 2018, 123, 28-42.	0.5	24
96	The Clinical Application of RNA Sequencing in Genetic Diagnosis of Mendelian Disorders. Advances in Molecular Pathology, 2018, 1, 27-36.	0.2	1
97	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
98	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
99	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
100	OCR-Stats: Robust estimation and statistical testing of mitochondrial respiration activities using Seahorse XF Analyzer. PLoS ONE, 2018, 13, e0199938.	1.1	55
101	PRUNE1 Deficiency: Expanding the Clinical and Genetic Spectrum. Neuropediatrics, 2018, 49, 330-338.	0.3	11
102	Advancing genomic approaches to the molecular diagnosis of mitochondrial disease. Essays in Biochemistry, 2018, 62, 399-408.	2.1	51
103	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
104	DNA methylation age is associated with an altered hemostatic profile in a multiethnic meta-analysis. Blood, 2018, 132, 1842-1850.	0.6	16
105	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
106	Mutations in PPCS, Encoding Phosphopantothenoylcysteine Synthetase, Cause Autosomal-Recessive Dilated Cardiomyopathy. American Journal of Human Genetics, 2018, 102, 1018-1030.	2.6	42
107	HTRA2 Defect: A Recognizable Inborn Error of Metabolism with 3-Methylglutaconic Aciduria as Discriminating Feature Characterized by Neonatal Movement Disorder and Epilepsyâ€"Report of 11 Patients. Neuropediatrics, 2018, 49, 373-378.	0.3	21
108	Breast cancer patients suggestive of Li-Fraumeni syndrome: mutational spectrum, candidate genes, and unexplained heredity. Breast Cancer Research, 2018, 20, 87.	2.2	9

#	Article	IF	Citations
109	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
110	A Homozygous Splice Site Mutation in SLC25A42, Encoding the Mitochondrial Transporter of Coenzyme A, Causes Metabolic Crises and Epileptic Encephalopathy. JIMD Reports, 2018, 44, 1-7.	0.7	15
111	Biallelic Mutations in DNAJC12 Cause Hyperphenylalaninemia, Dystonia, and Intellectual Disability. American Journal of Human Genetics, 2017, 100, 257-266.	2.6	127
112	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
113	Clinical Features, Molecular Heterogeneity, and Prognostic Implications in YARS2-Related Mitochondrial Myopathy. JAMA Neurology, 2017, 74, 686.	4.5	41
114	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
115	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
116	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
117	Genetic diagnosis of Mendelian disorders via RNA sequencing. Nature Communications, 2017, 8, 15824.	5.8	432
118	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
119	Analysis of Mitochondrial RNA-Processing Defects in Patient-Derived Tissues by qRT-PCR and RNAseq. Methods in Molecular Biology, 2017, 1567, 379-390.	0.4	8
120	CADmutations and uridine-responsive epileptic encephalopathy. Brain, 2017, 140, 279-286.	3.7	106
121	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
122	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
123	Treatable mitochondrial diseases: cofactor metabolism and beyond. Brain, 2017, 140, e11-e11.	3.7	57
124	Epigenome-wide association study of body mass index, and the adverse outcomes of adiposity. Nature, 2017, 541, 81-86.	13.7	743
125	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
126	Novel GFM2 variants associated with early-onset neurological presentations of mitochondrial disease and impaired expression of OXPHOS subunits. Neurogenetics, 2017, 18, 227-235.	0.7	10

#	Article	IF	Citations
127	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
128	Biallelic variants inWARS2encoding mitochondrial tryptophanyl-tRNA synthase in six individuals with mitochondrial encephalopathy. Human Mutation, 2017, 38, 1786-1795.	1.1	24
129	Molecular and clinical spectra of FBXL4 deficiency. Human Mutation, 2017, 38, 1649-1659.	1.1	41
130	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
131	Transcriptome-Wide Analysis Identifies Novel Associations With Blood Pressure. Hypertension, 2017, 70, 743-750.	1.3	34
132	LYRM7 - associated complex III deficiency: A clinical, molecular genetic, MR tomographic, and biochemical study. Mitochondrion, 2017, 37, 55-61.	1.6	20
133	Rare causes of early-onset dystonia-parkinsonism with cognitive impairment: a de novo PSEN-1 mutation. Neurogenetics, 2017, 18, 175-178.	0.7	23
134	ACSL4 dictates ferroptosis sensitivity by shaping cellular lipid composition. Nature Chemical Biology, 2017, 13, 91-98.	3.9	2,069
135	Neonatal encephalocardiomyopathy caused by mutations in VARS2. Metabolic Brain Disease, 2017, 32, 267-270.	1.4	26
136	Characterization of a Leber's hereditary optic neuropathy (LHON) family harboring two primary LHON mutations m.11778G > A and m.14484T > C of the mitochondrial DNA. Mitochondrion, 2017, 36, 15-20.	1.6	23
137	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
138	High Symmetry of Visual Acuity and Visual Fields in RPGR-Linked Retinitis Pigmentosa. , 2017, 58, 4457.		21
139	Epigenetic Signatures at AQP3 and SOCS3 Engage in Low-Grade Inflammation across Different Tissues. PLoS ONE, 2016, 11, e0166015.	1.1	14
140	Association between DNA Methylation in Whole Blood and Measures of Glucose Metabolism: KORA F4 Study. PLoS ONE, 2016, 11, e0152314.	1.1	81
141	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
142	Mitochondrial replacement approaches: challenges for clinical implementation. Genome Medicine, 2016, 8, 126.	3.6	21
143	DNA methylation signatures of chronic low-grade inflammation are associated with complex diseases. Genome Biology, 2016, 17, 255.	3.8	251
144	Genetic cause and prevalence of hydroxyprolinemia. Journal of Inherited Metabolic Disease, 2016, 39, 625-632.	1.7	17

#	Article	IF	Citations
145	Severe respiratory complex III defect prevents liver adaptation to prolonged fasting. Journal of Hepatology, 2016, 65, 377-385.	1.8	25
146	Mitochondrial Protein Interaction Mapping Identifies Regulators of Respiratory Chain Function. Molecular Cell, 2016, 63, 621-632.	4.5	241
147	The many faces of paediatric mitochondrial disease on neuroimaging. Child's Nervous System, 2016, 32, 2077-2083.	0.6	20
148	A Whole-Blood Transcriptome Meta-Analysis Identifies Gene Expression Signatures of Cigarette Smoking. Human Molecular Genetics, 2016, 25, ddw288.	1.4	76
149	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
150	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
151	Sudden Cardiac Death Due to Deficiency of the Mitochondrial Inorganic Pyrophosphatase PPA2. American Journal of Human Genetics, 2016, 99, 674-682.	2.6	48
152	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
153	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
154	Deficient methylation and formylation of mt-tRNAMet wobble cytosine in a patient carrying mutations in NSUN3. Nature Communications, 2016, 7, 12039.	5.8	178
155	Mutations in SLC39A14 disrupt manganese homeostasis and cause childhood-onset parkinsonismâ€“dystonia. Nature Communications, 2016, 7, 11601.	5.8	233
156	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
157	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
158	Bezafibrate Improves Insulin Sensitivity and Metabolic Flexibility in STZ-Induced Diabetic Mice. Diabetes, 2016, 65, 2540-2552.	0.3	35
159	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
160	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
161	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). Autophagy, 2016, 12, 1-222.	4.3	4,701
162	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

#	Article	IF	Citations
163	EARS2 mutations cause fatal neonatal lactic acidosis, recurrent hypoglycemia and agenesis of corpus callosum. Metabolic Brain Disease, 2016, 31, 717-721.	1.4	15
164	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
165	Analyzing Illumina Gene Expression Microarray Data Obtained From Human Whole Blood Cell and Blood Monocyte Samples. Methods in Molecular Biology, 2016, 1368, 85-97.	0.4	2
166	Human thioredoxin 2 deficiency impairs mitochondrial redox homeostasis and causes early-onset neurodegeneration. Brain, 2016, 139, 346-354.	3.7	86
167	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
168	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
169	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
170	Clinical, biochemical, and genetic spectrum of seven patients with NFU1 deficiency. Frontiers in Genetics, 2015, 06, 123.	1.1	81
171	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
172	Mitochondrial GWA Analysis of Lipid Profile Identifies Genetic Variants to Be Associated with HDL Cholesterol and Triglyceride Levels. PLoS ONE, 2015, 10, e0126294.	1.1	14
173	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
174	Mutations in TTC19: expanding the molecular, clinical and biochemical phenotype. Orphanet Journal of Rare Diseases, 2015, 10, 40.	1.2	25
175	LRPPRCmutations cause early-onset multisystem mitochondrial disease outside of the French-Canadian population. Brain, 2015, 138, 3503-3519.	3.7	81
176	Atypical Clinical Presentations of TAZ Mutations: An Underdiagnosed Cause of Growth Retardation?. JIMD Reports, 2015, 29, 89-93.	0.7	8
177	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
178	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
179	The spectrum of pyruvate oxidation defects in the diagnosis of mitochondrial disorders. Journal of Inherited Metabolic Disease, 2015, 38, 391-403.	1.7	44
180	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

#	Article	IF	Citations
181	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
182	DNA Methylation of Lipid-Related Genes Affects Blood Lipid Levels. Circulation: Cardiovascular Genetics, 2015, 8, 334-342.	5.1	151
183	Cell Specific eQTL Analysis without Sorting Cells. PLoS Genetics, 2015, 11, e1005223.	1.5	115
184	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
185	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
186	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
187	A Meta-analysis of Gene Expression Signatures of Blood Pressure and Hypertension. PLoS Genetics, 2015, 11, e1005035.	1.5	107
188	MRPL44 mutations cause a slowly progressive multisystem disease with childhood-onset hypertrophic cardiomyopathy. Neurogenetics, 2015, 16, 319-323.	0.7	44
189	Spectrum of combined respiratory chain defects. Journal of Inherited Metabolic Disease, 2015, 38, 629-640.	1.7	102
190	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
191	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
192	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
193	The transcriptional landscape of age in human peripheral blood. Nature Communications, 2015, 6, 8570.	5.8	533
194	MRPS22 mutation causes fatal neonatal lactic acidosis with brain and heart abnormalities. Neurogenetics, 2015, 16, 237-240.	0.7	34
195	Characterization of whole-genome autosomal differences of DNA methylation between men and women. Epigenetics and Chromatin, 2015, 8, 43.	1.8	176
196	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
197	The Human Blood Metabolome-Transcriptome Interface. PLoS Genetics, 2015, 11, e1005274.	1.5	99
198	Mapping the Genetic Architecture of Gene Regulation in Whole Blood. PLoS ONE, 2014, 9, e93844.	1.1	31

#	Article	IF	Citations
199	MTO1-Deficient Mouse Model Mirrors the Human Phenotype Showing Complex I Defect and Cardiomyopathy. PLoS ONE, 2014, 9, e114918.	1.1	17
200	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
201	Identification of Novel Genetic Loci Associated with Thyroid Peroxidase Antibodies and Clinical Thyroid Disease. PLoS Genetics, 2014, 10, e1004123.	1.5	150
202	Infantile Leigh-like syndrome caused by SLC19A3 mutations is a treatable disease. Brain, 2014, 137, e295-e295.	3.7	36
203	Genetic Determinants of Circulating Interleukin-1 Receptor Antagonist Levels and Their Association With Glycemic Traits. Diabetes, 2014, 63, 4343-4359.	0.3	40
204	Absence of BiP Co-chaperone DNAJC3 Causes Diabetes Mellitus and Multisystemic Neurodegeneration. American Journal of Human Genetics, 2014, 95, 689-697.	2.6	100
205	Mutations inSDHDlead to autosomal recessive encephalomyopathy and isolated mitochondrial complex II deficiency. Journal of Medical Genetics, 2014, 51, 170-175.	1.5	75
206	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
207	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
208	VARS2andTARS2Mutations in Patients with Mitochondrial Encephalomyopathies. Human Mutation, 2014, 35, 983-989.	1.1	86
209	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
210	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
211	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
212	Novel (ovario) leukodystrophy related to AARS2 mutations. Neurology, 2014, 82, 2063-2071.	1.5	172
213	Impairment of Drosophila Orthologs of the Human Orphan Protein C19orf12 Induces Bang Sensitivity and Neurodegeneration. PLoS ONE, 2014, 9, e89439.	1.1	28
214	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
215	Mutations in FBXL4, Encoding a Mitochondrial Protein, Cause Early-Onset Mitochondrial Encephalomyopathy. American Journal of Human Genetics, 2013, 93, 482-495.	2.6	138
216	ELAC2 Mutations Cause a Mitochondrial RNA Processing Defect Associated with Hypertrophic Cardiomyopathy. American Journal of Human Genetics, 2013, 93, 211-223.	2.6	127

#	Article	IF	Citations
217	Systematic identification of trans eQTLs as putative drivers of known disease associations. Nature Genetics, 2013, 45, 1238-1243.	9.4	1,544
218	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
219	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
220	Macrocytic Anemia and Mitochondriopathy Resulting from a Defect in Sideroflexin 4. American Journal of Human Genetics, 2013, 93, 906-914.	2.6	58
221	Mitochondrial membrane protein associated neurodegenration: A novel variant of neurodegeneration with brain iron accumulation. Movement Disorders, 2013, 28, 224-227.	2.2	162
222	Loss-of-function mutations in MGME1 impair mtDNA replication and cause multisystemic mitochondrial disease. Nature Genetics, 2013, 45, 214-219.	9.4	198
223	Beta-propeller protein-associated neurodegeneration: a new X-linked dominant disorder with brain iron accumulation. Brain, 2013, 136, 1708-1717.	3.7	203
224	BPAN. International Review of Neurobiology, 2013, 110, 85-90.	0.9	41
225	ADCK4 mutations promote steroid-resistant nephrotic syndrome through CoQ10 biosynthesis disruption. Journal of Clinical Investigation, 2013, 123, 5179-5189.	3.9	275
226	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
227	Leukoencephalopathy with thalamus and brainstem involvement and high lactate â€˜LTBLâ€™ caused by EARS2 mutations. Brain, 2012, 135, 1387-1394.	3.7	187
228	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
229	Molecular diagnosis in mitochondrial complex I deficiency using exome sequencing. Journal of Medical Genetics, 2012, 49, 277-283.	1.5	182
230	DHTKD1 Mutations Cause 2-Aminoadipic and 2-Oxoadipic Aciduria. American Journal of Human Genetics, 2012, 91, 1082-1087.	2.6	94
231	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
232	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
233	Lack of the Mitochondrial Protein Acylglycerol Kinase Causes Sengers Syndrome. American Journal of Human Genetics, 2012, 90, 314-320.	2.6	192
234	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

#	Article	IF	Citations
235	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
236	Cellular rescue-assay aids verification of causative DNA-variants in mitochondrial complex I deficiency. Molecular Genetics and Metabolism, 2011, 103, 161-166.	0.5	23
237	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
238	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
239	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
240	COQ6 mutations in human patients produce nephrotic syndrome with sensorineural deafness. Journal of Clinical Investigation, 2011, 121, 2013-2024.	3.9	343
241	Exome sequencing identifies ACAD9 mutations as a cause of complex I deficiency. Nature Genetics, 2010, 42, 1131-1134.	9.4	234
242	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
243	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
244	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
245	MitoP2: An Integrative Tool for the Analysis of the Mitochondrial Proteome. Molecular Biotechnology, 2008, 40, 306-315.	1.3	69
246	SLC2A9 influences uric acid concentrations with pronounced sex-specific effects. Nature Genetics, 2008, 40, 430-436.	9.4	363
247	A Population-Based Epidemiological and Genetic Study of X-Linked Retinitis Pigmentosa. , 2007, 48, 4012.		36
248	MitoP2, an Integrated Database for Mitochondrial Proteins. Methods in Molecular Biology, 2007, 372, 573-586.	0.4	11
249	Assessing Systems Properties of Yeast Mitochondria through an Interaction Map of the Organelle. PLoS Genetics, 2006, 2, e170.	1.5	67
250	Integrative Analysis of the Mitochondrial Proteome in Yeast. PLoS Biology, 2004, 2, e160.	2.6	181
251	Systematic screen for human disease genes in yeast. Nature Genetics, 2002, 31, 400-404.	9.4	503