Holger Prokisch

List of Publications by Citations

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253 papers	18,761 citations	63 h-index	132 g-index
274 ext. papers	23,277 ext. citations	8.2 avg, IF	5.97 L-index

#	Paper	IF	Citations
253	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). *Autophagy*, **2016**, 12, 1-222	10.2	3838
252	Systematic identification of trans eQTLs as putative drivers of known disease associations. *Nature Genetics*, **2013**, 45, 1238-1243	36.3	1244
251	ACSL4 dictates ferroptosis sensitivity by shaping cellular lipid composition. *Nature Chemical Biology* , **2017**, 13, 91-98	11.7	908
250	Epigenome-wide association study of body mass index, and the adverse outcomes of adiposity. *Nature*, **2017**, 541, 81-86	50.4	511
249	A genome-wide meta-analysis identifies 22 loci associated with eight hematological parameters in the HaemGen consortium. *Nature Genetics*, **2009**, 41, 1182-90	36.3	433
248	Systematic screen for human disease genes in yeast. *Nature Genetics*, **2002**, 31, 400-4	36.3	431
247	The transcriptional landscape of age in human peripheral blood. *Nature Communications*, **2015**, 6, 8570	17.4	335
246	SLC2A9 influences uric acid concentrations with pronounced sex-specific effects. *Nature Genetics*, **2008**, 40, 430-6	36.3	317
245	COQ6 mutations in human patients produce nephrotic syndrome with sensorineural deafness. *Journal of Clinical Investigation*, **2011**, 121, 2013-24	15.9	292
244	Genetic diagnosis of Mendelian disorders via RNA sequencing. *Nature Communications*, **2017**, 8, 15824	17.4	277
243	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	18.1	277
242	Exome sequencing reveals de novo WDR45 mutations causing a phenotypically distinct, X-linked dominant form of NBIA. *American Journal of Human Genetics*, **2012**, 91, 1144-9	11	268
241	ADCK4 mutations promote steroid-resistant nephrotic syndrome through CoQ10 biosynthesis disruption. *Journal of Clinical Investigation*, **2013**, 123, 5179-89	15.9	231
240	Exome sequencing identifies ACAD9 mutations as a cause of complex I deficiency. *Nature Genetics*, **2010**, 42, 1131-4	36.3	210
239	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, 543-50	11	182
238	Unraveling the polygenic architecture of complex traits using blood eQTL metaanalysis		175
237	DNA methylation signatures of chronic low-grade inflammation are associated with complex diseases. *Genome Biology*, **2016**, 17, 255	18.3	171

236	-Propeller protein-associated neurodegeneration: a new X-linked dominant disorder with brain iron accumulation. *Brain*, **2013**, 136, 1708-17	11.2	167
235	Loss-of-function mutations in MGME1 impair mtDNA replication and cause multisystemic mitochondrial disease. *Nature Genetics*, **2013**, 45, 214-9	36.3	166
234	Leukoencephalopathy with thalamus and brainstem involvement and high lactate TBLCaused by EARS2 mutations. *Brain*, **2012**, 135, 1387-94	11.2	165
233	Integrative analysis of the mitochondrial proteome in yeast. *PLoS Biology*, **2004**, 2, e160	9.7	165
232	Mitochondrial Protein Interaction Mapping Identifies Regulators of Respiratory Chain Function. *Molecular Cell*, **2016**, 63, 621-632	17.6	163
231	Mutations in SLC39A14 disrupt manganese homeostasis and cause childhood-onset parkinsonism-dystonia. *Nature Communications*, **2016**, 7, 11601	17.4	160
230	Lack of the mitochondrial protein acylglycerol kinase causes Sengers syndrome. *American Journal of Human Genetics*, **2012**, 90, 314-20	11	160
229	Molecular diagnosis in mitochondrial complex I deficiency using exome sequencing. *Journal of Medical Genetics*, **2012**, 49, 277-83	5.8	145
228	Novel (ovario) leukodystrophy related to AARS2 mutations. *Neurology*, **2014**, 82, 2063-71	6.5	142
227	Mutations of the mitochondrial-tRNA modifier MTO1 cause hypertrophic cardiomyopathy and lactic acidosis. *American Journal of Human Genetics*, **2012**, 90, 1079-87	11	140
226	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	11	137
225	Deficient methylation and formylation of mt-tRNA(Met) wobble cytosine in a patient carrying mutations in NSUN3. *Nature Communications*, **2016**, 7, 12039	17.4	124
224	DNA methylation of lipid-related genes affects blood lipid levels. *Circulation: Cardiovascular Genetics*, **2015**, 8, 334-42		122
223	Identification of novel genetic Loci associated with thyroid peroxidase antibodies and clinical thyroid disease. *PLoS Genetics*, **2014**, 10, e1004123	6	122
222	Characterization of whole-genome autosomal differences of DNA methylation between men and women. *Epigenetics and Chromatin*, **2015**, 8, 43	5.8	111
221	Mutations in FBXL4, encoding a mitochondrial protein, cause early-onset mitochondrial encephalomyopathy. *American Journal of Human Genetics*, **2013**, 93, 482-95	11	106
220	ELAC2 mutations cause a mitochondrial RNA processing defect associated with hypertrophic cardiomyopathy. *American Journal of Human Genetics*, **2013**, 93, 211-23	11	104
219	Expression analysis of dopaminergic neurons in Parkinson disease and aging links transcriptional dysregulation of energy metabolism to cell death. *Acta Neuropathologica*, **2011**, 122, 75-86	14.3	102

218	Riboflavin-responsive oxidative phosphorylation complex I deficiency caused by defective ACAD9: new function for an old gene. *Brain*, **2011**, 134, 210-9	11.2	100
217	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	11	97
216	Mutations in GTPBP3 cause a mitochondrial translation defect associated with hypertrophic cardiomyopathy, lactic acidosis, and encephalopathy. *American Journal of Human Genetics*, **2014**, 95, 708-20	11	95
215	A genome-wide association study identifies three loci associated with mean platelet volume. *American Journal of Human Genetics*, **2009**, 84, 66-71	11	94
214	Thiamine pyrophosphokinase deficiency in encephalopathic children with defects in the pyruvate oxidation pathway. *American Journal of Human Genetics*, **2011**, 89, 806-12	11	92
213	CLPB mutations cause 3-methylglutaconic aciduria, progressive brain atrophy, intellectual disability, congenital neutropenia, cataracts, movement disorder. *American Journal of Human Genetics*, **2015**, 96, 245-57	11	84
212	Biallelic Mutations in NBAS Cause Recurrent Acute Liver Failure with Onset in Infancy. *American Journal of Human Genetics*, **2015**, 97, 163-9	11	83
211	A meta-analysis of gene expression signatures of blood pressure and hypertension. *PLoS Genetics*, **2015**, 11, e1005035	6	83
210	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, 270-8	5.8	83
209	Biallelic Mutations in DNAJC12 Cause Hyperphenylalaninemia, Dystonia, and Intellectual Disability. *American Journal of Human Genetics*, **2017**, 100, 257-266	11	81
208	Cell Specific eQTL Analysis without Sorting Cells. *PLoS Genetics*, **2015**, 11, e1005223	6	81
207	Absence of BiP co-chaperone DNAJC3 causes diabetes mellitus and multisystemic neurodegeneration. *American Journal of Human Genetics*, **2014**, 95, 689-97	11	79
206	Spectrum of combined respiratory chain defects. *Journal of Inherited Metabolic Disease*, **2015**, 38, 629-40	5.4	78
205	CAD mutations and uridine-responsive epileptic encephalopathy. *Brain*, **2017**, 140, 279-286	11.2	77
204	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	4.3	77
203	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	11	69
202	Homozygous missense mutation in BOLA3 causes multiple mitochondrial dysfunctions syndrome in two siblings. *Journal of Inherited Metabolic Disease*, **2013**, 36, 55-62	5.4	69
201	Deficiency of ECHS1 causes mitochondrial encephalopathy with cardiac involvement. *Annals of Clinical and Translational Neurology*, **2015**, 2, 492-509	5.3	69

200	Recurrent acute liver failure due to NBAS deficiency: phenotypic spectrum, disease mechanisms, and therapeutic concepts. *Journal of Inherited Metabolic Disease*, **2016**, 39, 3-16	5.4	68
199	Mutation screening of 75 candidate genes in 152 complex I deficiency cases identifies pathogenic variants in 16 genes including NDUFB9. *Journal of Medical Genetics*, **2012**, 49, 83-9	5.8	68
198	DHTKD1 mutations cause 2-aminoadipic and 2-oxoadipic aciduria. *American Journal of Human Genetics*, **2012**, 91, 1082-7	11	67
197	VARS2 and TARS2 mutations in patients with mitochondrial encephalomyopathies. *Human Mutation* , **2014**, 35, 983-9	4.7	66
196	COQ4 mutations cause a broad spectrum of mitochondrial disorders associated with CoQ10 deficiency. *American Journal of Human Genetics*, **2015**, 96, 309-17	11	66
195	MitoP2: an integrative tool for the analysis of the mitochondrial proteome. *Molecular Biotechnology* , **2008**, 40, 306-15	3	65
194	The Human Blood Metabolome-Transcriptome Interface. *PLoS Genetics*, **2015**, 11, e1005274	6	65
193	Genetics of mitochondrial diseases: Identifying mutations to help diagnosis. *EBioMedicine*, **2020**, 56, 102784	11.4	64
192	Human thioredoxin 2 deficiency impairs mitochondrial redox homeostasis and causes early-onset neurodegeneration. *Brain*, **2016**, 139, 346-54	11.2	64
191	Clinical, biochemical, and genetic spectrum of seven patients with NFU1 deficiency. *Frontiers in Genetics*, **2015**, 6, 123	4.5	64
190	LRPPRC mutations cause early-onset multisystem mitochondrial disease outside of the French-Canadian population. *Brain*, **2015**, 138, 3503-19	11.2	63
189	Assessing systems properties of yeast mitochondria through an interaction map of the organelle. *PLoS Genetics*, **2006**, 2, e170	6	63
188	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, 319-28	11	62
187	MTO1 mutations are associated with hypertrophic cardiomyopathy and lactic acidosis and cause respiratory chain deficiency in humans and yeast. *Human Mutation*, **2013**, 34, 1501-9	4.7	62
186	Recessive mutations in VPS13D cause childhood onset movement disorders. *Annals of Neurology*, **2018**, 83, 1089-1095	9.4	61
185	Mitochondria: Much ado about nothing? How dangerous is reactive oxygen species production?. *International Journal of Biochemistry and Cell Biology*, **2015**, 63, 16-20	5.6	61
184	Impaired riboflavin transport due to missense mutations in SLC52A2 causes Brown-Vialetto-Van Laere syndrome. *Journal of Inherited Metabolic Disease*, **2012**, 35, 943-8	5.4	60
183	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	36.3	60

182	A whole-blood transcriptome meta-analysis identifies gene expression signatures of cigarette smoking. *Human Molecular Genetics*, **2016**, 25, 4611-4623	5.6	58
181	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	3.7	58
180	Mutations in SDHD lead to autosomal recessive encephalomyopathy and isolated mitochondrial complex II deficiency. *Journal of Medical Genetics*, **2014**, 51, 170-5	5.8	58
179	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, 388-96	3.7	56
178	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	11	54
177	Analyzing illumina gene expression microarray data from different tissues: methodological aspects of data analysis in the metaxpress consortium. *PLoS ONE*, **2012**, 7, e50938	3.7	54
176	Monogenic variants in dystonia: an exome-wide sequencing study. *Lancet Neurology, The*, **2020**, 19, 908-918	24.1	51
175	Association between DNA Methylation in Whole Blood and Measures of Glucose Metabolism: KORA F4 Study. *PLoS ONE*, **2016**, 11, e0152314	3.7	50
174	Bi-allelic Truncating Mutations in TANGO2 Cause Infancy-Onset Recurrent Metabolic Crises with Encephalocardiomyopathy. *American Journal of Human Genetics*, **2016**, 98, 358-62	11	49
173	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. *Journal of Clinical Investigation*, **2020**, 130, 108-125	15.9	49
172	Biallelic IARS Mutations Cause Growth Retardation with Prenatal Onset, Intellectual Disability, Muscular Hypotonia, and Infantile Hepatopathy. *American Journal of Human Genetics*, **2016**, 99, 414-22	11	48
171	Mutations in APOPT1, encoding a mitochondrial protein, cause cavitating leukoencephalopathy with cytochrome c oxidase deficiency. *American Journal of Human Genetics*, **2014**, 95, 315-25	11	48
170	CoQ deficiency causes disruption of mitochondrial sulfide oxidation, a new pathomechanism associated with this syndrome. *EMBO Molecular Medicine*, **2017**, 9, 78-95	12	47
169	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	11	47
168	Treatable mitochondrial diseases: cofactor metabolism and beyond. *Brain*, **2017**, 140, e11	11.2	46
167	Biallelic Mutations in TMEM126B Cause Severe Complex I Deficiency with a Variable Clinical Phenotype. *American Journal of Human Genetics*, **2016**, 99, 217-27	11	45
166	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	11	44
165	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, 3238-47	5.6	44

164	Macrocytic anemia and mitochondriopathy resulting from a defect in sideroflexin 4. *American Journal of Human Genetics*, **2013**, 93, 906-14	11	42
163	Advancing genomic approaches to the molecular diagnosis of mitochondrial disease. *Essays in Biochemistry*, **2018**, 62, 399-408	7.6	41
162	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. *Orphanet Journal of Rare Diseases*, **2018**, 13, 120	4.2	41
161	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 Ca□+. *Frontiers in Genetics*, **2015**, 6, 185	4.5	41
160	Assessing Mitochondrial Bioenergetics in Isolated Mitochondria from Various Mouse Tissues Using Seahorse XF96 Analyzer. *Methods in Molecular Biology*, **2017**, 1567, 217-230	1.4	40
159	Delineating -associated disease: From isolated neuropathy to early onset neurodegeneration. *Neurology: Genetics*, **2020**, 6, e393	3.8	40
158	Expanding the clinical and molecular spectrum of thiamine pyrophosphokinase deficiency: a treatable neurological disorder caused by TPK1 mutations. *Molecular Genetics and Metabolism*, **2014**, 113, 301-6	3.7	40
157	Single-cell expression profiling of dopaminergic neurons combined with association analysis identifies pyridoxal kinase as Parkinson□ disease gene. *Annals of Neurology*, **2009**, 66, 792-8	9.4	40
156	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	1.6	39
155	GWAS on longitudinal growth traits reveals different genetic factors influencing infant, child, and adult BMI. *Science Advances*, **2019**, 5, eaaw3095	14.3	39
154	MRPL44 mutations cause a slowly progressive multisystem disease with childhood-onset hypertrophic cardiomyopathy. *Neurogenetics*, **2015**, 16, 319-23	3	39
153	Impact of common regulatory single-nucleotide variants on gene expression profiles in whole blood. *European Journal of Human Genetics*, **2013**, 21, 48-54	5.3	39
152	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, 2247-66	5.6	39
151	Mutations in MDH2, Encoding a Krebs Cycle Enzyme, Cause Early-Onset Severe Encephalopathy. *American Journal of Human Genetics*, **2017**, 100, 151-159	11	38
150	Mitochondrial DNA mutation analysis from exome sequencing-A more holistic approach in diagnostics of suspected mitochondrial disease. *Journal of Inherited Metabolic Disease*, **2019**, 42, 909-917	5.4	38
149	OUTRIDER: A Statistical Method for Detecting Aberrantly Expressed Genes in RNA Sequencing Data. *American Journal of Human Genetics*, **2018**, 103, 907-917	11	37
148	The spectrum of pyruvate oxidation defects in the diagnosis of mitochondrial disorders. *Journal of Inherited Metabolic Disease*, **2015**, 38, 391-403	5.4	36
147	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, 905-14	5.4	35

146	Quantification and discovery of sequence determinants of protein-per-mRNA amount in 29 human tissues. *Molecular Systems Biology*, **2019**, 15, e8513	12.2	33
145	NDUFB8 Mutations Cause Mitochondrial Complex I Deficiency in Individuals with Leigh-like Encephalomyopathy. *American Journal of Human Genetics*, **2018**, 102, 460-467	11	33
144	Sudden Cardiac Death Due to Deficiency of the Mitochondrial Inorganic Pyrophosphatase PPA2. *American Journal of Human Genetics*, **2016**, 99, 674-682	11	33
143	Genetic determinants of circulating interleukin-1 receptor antagonist levels and their association with glycemic traits. *Diabetes*, **2014**, 63, 4343-59	0.9	32
142	BPAN: the only X-linked dominant NBIA disorder. *International Review of Neurobiology*, **2013**, 110, 85-90	4.4	32
141	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	11	32
140	Fatal neonatal encephalopathy and lactic acidosis caused by a homozygous loss-of-function variant in COQ9. *European Journal of Human Genetics*, **2016**, 24, 450-4	5.3	31
139	Clinical Features, Molecular Heterogeneity, and Prognostic Implications in YARS2-Related Mitochondrial Myopathy. *JAMA Neurology*, **2017**, 74, 686-694	17.2	31
138	Biallelic Mutations in LIPT2 Cause a Mitochondrial Lipoylation Defect Associated with Severe Neonatal Encephalopathy. *American Journal of Human Genetics*, **2017**, 101, 283-290	11	31
137	NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. *Brain*, **2019**, 142, 50-58	11.2	31
136	A truncating PET100 variant causing fatal infantile lactic acidosis and isolated cytochrome c oxidase deficiency. *European Journal of Human Genetics*, **2015**, 23, 935-9	5.3	30
135	Mitochondrial membrane protein associated neurodegenration: a novel variant of neurodegeneration with brain iron accumulation. *Movement Disorders*, **2013**, 28, 224-7	7	30
134	SCYL1 variants cause a syndrome with low γ-glutamyl-transferase cholestasis, acute liver failure, and neurodegeneration (CALFAN). *Genetics in Medicine*, **2018**, 20, 1255-1265	8.1	29
133	Mutations in PPCS, Encoding Phosphopantothenoylcysteine Synthetase, Cause Autosomal-Recessive Dilated Cardiomyopathy. *American Journal of Human Genetics*, **2018**, 102, 1018-1030	11	29
132	Molecular and clinical spectra of FBXL4 deficiency. *Human Mutation*, **2017**, 38, 1649-1659	4.7	29
131	Bainbridge-Ropers syndrome caused by loss-of-function variants in ASXL3: a recognizable condition. *European Journal of Human Genetics*, **2017**, 25, 183-191	5.3	28
130	MRPS22 mutation causes fatal neonatal lactic acidosis with brain and heart abnormalities. *Neurogenetics*, **2015**, 16, 237-40	3	28
129	Bezafibrate Improves Insulin Sensitivity and Metabolic Flexibility in STZ-Induced Diabetic Mice. *Diabetes*, **2016**, 65, 2540-52	0.9	28

128	Infantile Leigh-like syndrome caused by SLC19A3 mutations is a treatable disease. *Brain*, **2014**, 137, e295	11.2	28
127	OCR-Stats: Robust estimation and statistical testing of mitochondrial respiration activities using Seahorse XF Analyzer. *PLoS ONE*, **2018**, 13, e0199938	3.7	28
126	ncRNAs: New Players in Mitochondrial Health and Disease?. *Frontiers in Genetics*, **2020**, 11, 95	4.5	27
125	Combined Respiratory Chain Deficiency and Mutations in Neonatal Encephalomyopathy: Defective Supercomplex Assembly in Complex III Deficiencies. *Oxidative Medicine and Cellular Longevity*, **2017**, 2017, 7202589	6.7	27
124	Mapping the genetic architecture of gene regulation in whole blood. *PLoS ONE*, **2014**, 9, e93844	3.7	27
123	A population-based epidemiological and genetic study of X-linked retinitis pigmentosa. *Investigative Ophthalmology and Visual Science*, **2007**, 48, 4012-8		27
122	Impaired complex I repair causes recessive Leber hereditary optic neuropathy. *Journal of Clinical Investigation*, **2021**, 131,	15.9	27
121	"Transcriptomics": molecular diagnosis of inborn errors of metabolism via RNA-sequencing. *Journal of Inherited Metabolic Disease*, **2018**, 41, 525-532	5.4	26
120	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	4.8	25
119	Bi-allelic mutations in result in a neurodevelopmental disorder and have an impact on RAB11 in fibroblasts. *Journal of Medical Genetics*, **2018**, 55, 753-764	5.8	25
118	Pathogenic Bi-allelic Mutations in NDUFAF8 Cause Leigh Syndrome with an Isolated Complex I Deficiency. *American Journal of Human Genetics*, **2020**, 106, 92-101	11	25
117	The diagnosis of inborn errors of metabolism by an integrative "multi-omics" approach: A perspective encompassing genomics, transcriptomics, and proteomics. *Journal of Inherited Metabolic Disease*, **2020**, 43, 25-35	5.4	25
116	Bi-allelic ADPRHL2 Mutations Cause Neurodegeneration with Developmental Delay, Ataxia, and Axonal Neuropathy. *American Journal of Human Genetics*, **2018**, 103, 817-825	11	24
115	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	6.9	23
114	Cellular rescue-assay aids verification of causative DNA-variants in mitochondrial complex I deficiency. *Molecular Genetics and Metabolism*, **2011**, 103, 161-6	3.7	23
113	The Dimensions of Primary Mitochondrial Disorders. *Frontiers in Cell and Developmental Biology*, **2020**, 8, 600079	5.7	23
112	Severe respiratory complex III defect prevents liver adaptation to prolonged fasting. *Journal of Hepatology*, **2016**, 65, 377-85	13.4	23
111	Detection of aberrant splicing events in RNA-seq data using FRASER. *Nature Communications*, **2021**, 12, 529	17.4	23

110	Defining clinical subgroups and genotype-phenotype correlations in NBAS-associated disease across 110 patients. *Genetics in Medicine*, **2020**, 22, 610-621	8.1	22
109	Lifetime risk of autosomal recessive mitochondrial disorders calculated from genetic databases. *EBioMedicine*, **2020**, 54, 102730	8.8	22
108	Mutation in Gene Can Cause Syndromic Multisystem Autoimmune Disease With Acute Liver Failure. *Pediatrics*, **2019**, 143,	7.4	21
107	High incidence and variable clinical outcome of cardiac hypertrophy due to ACAD9 mutations in childhood. *European Journal of Human Genetics*, **2016**, 24, 1112-6	5.3	21
106	Transcriptome-Wide Analysis Identifies Novel Associations With Blood Pressure. *Hypertension*, **2017**, 70, 743-750	8.5	21
105	Neonatal encephalocardiomyopathy caused by mutations in VARS2. *Metabolic Brain Disease*, **2017**, 32, 267-270	3.9	21
104	Mutations in TTC19: expanding the molecular, clinical and biochemical phenotype. *Orphanet Journal of Rare Diseases*, **2015**, 10, 40	4.2	21
103	Impairment of Drosophila orthologs of the human orphan protein C19orf12 induces bang sensitivity and neurodegeneration. *PLoS ONE*, **2014**, 9, e89439	3.7	21
102	A recurrent mitochondrial p.Trp22Arg NDUFB3 variant causes a distinctive facial appearance, short stature and a mild biochemical and clinical phenotype. *Journal of Medical Genetics*, **2016**, 53, 634-41	5.8	20
101	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	11	20
100	Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults. *JAMA Neurology*, **2018**, 75, 105-113	17.2	20
99	Identification of Disease-Causing Mutations by Functional Complementation of Patient-Derived Fibroblast Cell Lines. *Methods in Molecular Biology*, **2017**, 1567, 391-406	1.4	19
98	Bi-Allelic UQCRFS1 Variants Are Associated with Mitochondrial Complex III Deficiency, Cardiomyopathy, and Alopecia Totalis. *American Journal of Human Genetics*, **2020**, 106, 102-111	11	19
97	RINT1 Bi-allelic Variations Cause Infantile-Onset Recurrent Acute Liver Failure and Skeletal Abnormalities. *American Journal of Human Genetics*, **2019**, 105, 108-121	11	18
96	Characterization of a Leber 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	4.9	18
95	The genotypic and phenotypic spectrum of MTO1 deficiency. *Molecular Genetics and Metabolism*, **2018**, 123, 28-42	3.7	18
94	Biallelic variants in the transcription factor PAX7 are a new genetic cause of myopathy. *Genetics in Medicine*, **2019**, 21, 2521-2531	8.1	17
93	Mutations in ELAC2 associated with hypertrophic cardiomyopathy impair mitochondrial tRNA 3′end processing. *Human Mutation*, **2019**, 40, 1731-1748	4.7	17

92	Association of Methylation Signals With Incident Coronary Heart Disease in an Epigenome-Wide Assessment of Circulating Tumor Necrosis Factor . *JAMA Cardiology*, **2018**, 3, 463-472	16.2	17
91	LYRM7 - associated complex III deficiency: A clinical, molecular genetic, MR tomographic, and biochemical study. *Mitochondrion*, **2017**, 37, 55-61	4.9	17
90	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	3.7	16
89	Detection of aberrant gene expression events in RNA sequencing data. *Nature Protocols*, **2021**, 16, 1276-1296	13.1	16
88	Detection of 6-demethoxyubiquinone in CoQ deficiency disorders: Insights into enzyme interactions and identification of potential therapeutics. *Molecular Genetics and Metabolism*, **2017**, 121, 216-223	3.7	15
87	Clinical, biochemical, and genetic features associated with VARS2-related mitochondrial disease. *Human Mutation*, **2018**, 39, 563-578	4.7	15
86	Biallelic variants in WARS2 encoding mitochondrial tryptophanyl-tRNA synthase in six individuals with mitochondrial encephalopathy. *Human Mutation*, **2017**, 38, 1786-1795	4.7	15
85	MTO1-deficient mouse model mirrors the human phenotype showing complex I defect and cardiomyopathy. *PLoS ONE*, **2014**, 9, e114918	3.7	15
84	Mitochondrial replacement approaches: challenges for clinical implementation. *Genome Medicine*, **2016**, 8, 126	14.4	15
83	NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. *Brain*, **2020**, 143, e8	11.2	14
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