Holger Prokisch

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	Leigh syndrome: a study of 209 patients at the Beijing Children© Hospital.. *Annals of Neurology*, **2022**,	9.4	2
252	Genetic variation influencing DNA methylation provides insights into molecular mechanisms regulating genomic function.. *Nature Genetics*, **2022**,	36.3	6
251	Genomic Approaches for the Diagnosis of Inborn Errors of Metabolism **2022**, 147-162		
250	RNA sequencing role and application in clinical diagnostic.. *Pediatric Investigation*, **2022**, 6, 29-35	1.3	1
249	Clinical implementation of RNA sequencing for Mendelian disease diagnostics.. *Genome Medicine*, **2022**, 14, 38	14.4	4
248	AOPEP variants as a novel cause of recessive dystonia: Generalized dystonia and dystonia-parkinsonism.. *Parkinsonism and Related Disorders*, **2022**, 97, 52-56	3.6	0
247	Population-based screening in children for early diagnosis and treatment of familial hypercholesterolemia: design of the VRONI study. *Medizinische Genetik*, **2022**, 34, 41-51	0.5	
246	Variants in mitochondrial ATP synthase cause variable neurologic phenotypes.. *Annals of Neurology*, **2021**,	9.4	1
245	Characterising a homozygous two-exon deletion in UQCRH: comparing human and mouse phenotypes. *EMBO Molecular Medicine*, **2021**, 13, e14397	12	0
244	Mitochondrial Disorders. *Deutsches Ärzteblatt International*, **2021**,	2.5	2
243	ATP synthase deficiency due to m.8528T>C mutation□ a novel cause of severe neonatal hyperammonemia requiring hemodialysis. *Journal of Pediatric Endocrinology and Metabolism*, **2021**, 34, 389-393	1.6	
242	Identification of a novel m.3955G□>□A variant in MT-ND1 associated with Leigh syndrome. *Mitochondrion*, **2021**, 62, 13-23	4.9	2
241	Multi-Omics Approach to Mitochondrial DNA Damage in Human Muscle Fibers. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	1
240	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	7.7	3
239	The genetics of mitochondrial disease: dissecting mitochondrial pathology using multi-omic pipelines. *Journal of Pathology*, **2021**, 254, 430-442	9.4	13
238	Novel NDUFA12 variants are associated with isolated complex I defect and variable clinical manifestation. *Human Mutation*, **2021**, 42, 699-710	4.7	4
237	Genetic basis of mitochondrial diseases. *FEBS Letters*, **2021**, 595, 1132-1158	3.8	12

236	Impaired complex I repair causes recessive Leber hereditary optic neuropathy. *Journal of Clinical Investigation*, **2021**, 131,	15.9	27
235	Whole genome and exome sequencing identify mutations as a new cause of progressive cavitating leukoencephalopathy. *Journal of Medical Genetics*, **2021**,	5.8	1
234	Identification of a Novel Variant in Causing MELAS. *Frontiers in Genetics*, **2021**, 12, 638749	4.5	0
233	A comprehensive phenotypic characterization of a whole-body Wdr45 knock-out mouse. *Mammalian Genome*, **2021**, 32, 332-349	3.2	1
232	Aberrant activity of mitochondrial NCLX is linked to impaired synaptic transmission and is associated with mental retardation. *Communications Biology*, **2021**, 4, 666	6.7	4
231	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	2.6	2
230	Myopathic mitochondrial DNA depletion syndrome associated with biallelic variants in LIG3. *Brain*, **2021**, 144, e74	11.2	2
229	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	5.6	9
228	Biallelic -Variants Leading to Developmental Regression With Progressive Spasticity and Brain Atrophy in a Chinese Patient. *Frontiers in Genetics*, **2021**, 12, 685035	4.5	1
227	Detection of aberrant splicing events in RNA-seq data using FRASER. *Nature Communications*, **2021**, 12, 529	17.4	23
226	Pediatric Leigh Syndrome: Neuroimaging Features and Genetic Correlations. *Annals of Neurology*, **2021**, 89, 629-631	9.4	3
225	X-Linked Retinitis Pigmentosa Caused by Non-Canonical Splice Site Variants in. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	3
224	Detection of aberrant gene expression events in RNA sequencing data. *Nature Protocols*, **2021**, 16, 1276-1296	18.2	16
223	DNA methylation and lipid metabolism: an EWAS of 226 metabolic measures. *Clinical Epigenetics*, **2021**, 13, 7	7.7	11
222	Expanding the clinical and genetic spectrum of FDXR deficiency by functional validation of variants of uncertain significance. *Human Mutation*, **2021**, 42, 310-319	4.7	4
221	Muscular and Molecular Pathology Associated with SPATA5 Deficiency in a Child with EHLMRS. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	0
220	NBAS Variants Are Associated with Quantitative and Qualitative NK and B Cell Deficiency. *Journal of Clinical Immunology*, **2021**, 41, 1781-1793	5.7	2
219	Pathogenic variants in MRPL44 cause infantile cardiomyopathy due to a mitochondrial translation defect. *Molecular Genetics and Metabolism*, **2021**, 133, 362-371	3.7	0

218	Prevalence and clinical prediction of mitochondrial disorders in a large neuropediatric cohort. *Clinical Genetics*, **2021**, 100, 766-770	4	1
217	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
216	A novel cryptic splice site mutation in as a cause of osteogenesis imperfecta. *Bone Reports*, **2021**, 15, 101110	2.6	1
215	Epigenome-wide association study of serum urate reveals insights into urate co-regulation and the SLC2A9 locus. *Nature Communications*, **2021**, 12, 7173	17.4	1
214	Meta-analyses identify DNA methylation associated with kidney function and damage. *Nature Communications*, **2021**, 12, 7174	17.4	0
213	Recurrent acute liver failure in alanyl-tRNA synthetase-1 (AARS1) deficiency. *Molecular Genetics and Metabolism Reports*, **2020**, 25, 100681	1.8	1
212	Genetics of mitochondrial diseases: Identifying mutations to help diagnosis. *EBioMedicine*, **2020**, 56, 102784	8.4	64
211	Delineating -associated disease: From isolated neuropathy to early onset neurodegeneration. *Neurology: Genetics*, **2020**, 6, e393	3.8	40
210	ncRNAs: New Players in Mitochondrial Health and Disease?. *Frontiers in Genetics*, **2020**, 11, 95	4.5	27
209	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. *Journal of Clinical Investigation*, **2020**, 130, 108-125	15.9	49
208	Blood DNA methylation sites predict death risk in a longitudinal study of 12, 300 individuals. *Aging*, **2020**, 12, 14092-14124	5.6	6
207	The Clinical Application of RNA Sequencing in Genetic Diagnosis of Mendelian Disorders. *Clinics in Laboratory Medicine*, **2020**, 40, 121-133	2.1	2
206	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
205	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
204	Defining clinical subgroups and genotype-phenotype correlations in NBAS-associated disease across 110 patients. *Genetics in Medicine*, **2020**, 22, 610-621	8.1	22
203	NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. *Brain*, **2020**, 143, e8	11.2	14
202	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	5.6	8
201	Monogenic variants in dystonia: an exome-wide sequencing study. *Lancet Neurology, The*, **2020**, 19, 908-918	24.1	51

200	Case Report: Rapid Treatment of Uridine-Responsive Epileptic Encephalopathy Caused by CAD Deficiency. *Frontiers in Pharmacology*, **2020**, 11, 608737	5.6	4
199	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	2.6	5
198	PKAN neurodegeneration and residual PANK2 activities in patient erythrocytes. *Annals of Clinical and Translational Neurology*, **2020**, 7, 1340-1351	5.3	4
197	Genotypic diversity and phenotypic spectrum of infantile liver failure syndrome type 1 due to variants in LARS1. *Genetics in Medicine*, **2020**, 22, 1863-1873	8.1	9
196	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	2.6	7
195	Mitochondrial Regulation of the 26S Proteasome. *Cell Reports*, **2020**, 32, 108059	10.6	13
194	Rescue of respiratory failure in pulmonary alveolar proteinosis due to pathogenic MARS1 variants. *Pediatric Pulmonology*, **2020**, 55, 3057-3066	3.5	8
193	The Dimensions of Primary Mitochondrial Disorders. *Frontiers in Cell and Developmental Biology*, **2020**, 8, 600079	5.7	23
192	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
191	Lifetime risk of autosomal recessive mitochondrial disorders calculated from genetic databases. *EBioMedicine*, **2020**, 54, 102730	8.8	22
190	GWAS on longitudinal growth traits reveals different genetic factors influencing infant, child, and adult BMI. *Science Advances*, **2019**, 5, eaaw3095	14.3	39
189	Biallelic variants in the transcription factor PAX7 are a new genetic cause of myopathy. *Genetics in Medicine*, **2019**, 21, 2521-2531	8.1	17
188	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
187	Mutations in ELAC2 associated with hypertrophic cardiomyopathy impair mitochondrial tRNA 3′end processing. *Human Mutation*, **2019**, 40, 1731-1748	4.7	17
186	Mitochondrial Disease Genetics **2019**, 41-62		
185	Quantification and discovery of sequence determinants of protein-per-mRNA amount in 29 human tissues. *Molecular Systems Biology*, **2019**, 15, e8513	12.2	33
184	The metabolic network coherence of human transcriptomes is associated with genetic variation at the cadherin 18 locus. *Human Genetics*, **2019**, 138, 375-388	6.3	4
183	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	5.3	3

182	Mutation in Gene Can Cause Syndromic Multisystem Autoimmune Disease With Acute Liver Failure. *Pediatrics*, **2019**, 143,	7.4	21
181	Molecular diagnostics of Mendelian disorders via combined DNA and RNA sequencing. *Medizinische Genetik*, **2019**, 31, 191-197	0.5	
180	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
179	Arabidopsis thaliana alternative dehydrogenases: a potential therapy for mitochondrial complex I deficiency? Perspectives and pitfalls. *Orphanet Journal of Rare Diseases*, **2019**, 14, 236	4.2	8
178	Genetic Basis of Mitochondrial Cardiomyopathy. *Cardiac and Vascular Biology*, **2019**, 93-139	0.2	1
177	NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. *Brain*, **2019**, 142, 50-58	11.2	31
176	A Homozygous Splice Site Mutation in SLC25A42, Encoding the Mitochondrial Transporter of Coenzyme A, Causes Metabolic Crises and Epileptic Encephalopathy. *JIMD Reports*, **2019**, 44, 1-7	1.9	12
175	Recessive mutations in VPS13D cause childhood onset movement disorders. *Annals of Neurology*, **2018**, 83, 1089-1095	9.4	61
174	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
173	"Transcriptomics": molecular diagnosis of inborn errors of metabolism via RNA-sequencing. *Journal of Inherited Metabolic Disease*, **2018**, 41, 525-532	5.4	26
172	SCYL1 variants cause a syndrome with low glutamyl-transferase cholestasis, acute liver failure, and neurodegeneration (CALFAN). *Genetics in Medicine*, **2018**, 20, 1255-1265	8.1	29
171	NDUFB8 Mutations Cause Mitochondrial Complex I Deficiency in Individuals with Leigh-like Encephalomyopathy. *American Journal of Human Genetics*, **2018**, 102, 460-467	11	33
170	Clinical, biochemical, and genetic features associated with VARS2-related mitochondrial disease. *Human Mutation*, **2018**, 39, 563-578	4.7	15
169	Severe ichthyosis in MPDU1-CDG. *Journal of Inherited Metabolic Disease*, **2018**, 41, 1293-1294	5.4	5
168	Biallelic Mutations in SLC1A2; an Additional Mode of Inheritance for SLC1A2-Related Epilepsy. *Neuropediatrics*, **2018**, 49, 59-62	1.6	9
167	Advancing genomic approaches to the molecular diagnosis of mitochondrial disease. *Essays in Biochemistry*, **2018**, 62, 399-408	7.6	41
166	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
165	DNA methylation age is associated with an altered hemostatic profile in a multiethnic meta-analysis. *Blood*, **2018**, 132, 1842-1850	2.2	11

164	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
163	Mutations in PPCS, Encoding Phosphopantothenoylcysteine Synthetase, Cause Autosomal-Recessive Dilated Cardiomyopathy. *American Journal of Human Genetics*, **2018**, 102, 1018-1030	11	29
162	HTRA2 Defect: A Recognizable Inborn Error of Metabolism with 3-Methylglutaconic Aciduria as Discriminating Feature Characterized by Neonatal Movement Disorder and Epilepsy-Report of 11 Patients. *Neuropediatrics*, **2018**, 49, 373-378	1.6	10
161	Breast cancer patients suggestive of Li-Fraumeni syndrome: mutational spectrum, candidate genes, and unexplained heredity. *Breast Cancer Research*, **2018**, 20, 87	8.3	4
160	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
159	Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults. *JAMA Neurology*, **2018**, 75, 105-113	17.2	20
158	The genotypic and phenotypic spectrum of MTO1 deficiency. *Molecular Genetics and Metabolism*, **2018**, 123, 28-42	3.7	18
157	The Clinical Application of RNA Sequencing in Genetic Diagnosis of Mendelian Disorders. *Advances in Molecular Pathology*, **2018**, 1, 27-36	0.3	1
156	OUTRIDER: A Statistical Method for Detecting Aberrantly Expressed Genes in RNA Sequencing Data. *American Journal of Human Genetics*, **2018**, 103, 907-917	11	37
155	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
154	Bi-allelic ADPRHL2 Mutations Cause Neurodegeneration with Developmental Delay, Ataxia, and Axonal Neuropathy. *American Journal of Human Genetics*, **2018**, 103, 817-825	11	24
153	OCR-Stats: Robust estimation and statistical testing of mitochondrial respiration activities using Seahorse XF Analyzer. *PLoS ONE*, **2018**, 13, e0199938	3.7	28
152	PRUNE1 Deficiency: Expanding the Clinical and Genetic Spectrum. *Neuropediatrics*, **2018**, 49, 330-338	1.6	8
151	Biallelic Mutations in DNAJC12 Cause Hyperphenylalaninemia, Dystonia, and Intellectual Disability. *American Journal of Human Genetics*, **2017**, 100, 257-266	11	81
150	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
149	Clinical Features, Molecular Heterogeneity, and Prognostic Implications in YARS2-Related Mitochondrial Myopathy. *JAMA Neurology*, **2017**, 74, 686-694	17.2	31
148	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
147	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

146	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
145	Genetic diagnosis of Mendelian disorders via RNA sequencing. *Nature Communications*, **2017**, 8, 15824	17.4	277
144	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
143	Analysis of Mitochondrial RNA-Processing Defects in Patient-Derived Tissues by qRT-PCR and RNAseq. *Methods in Molecular Biology*, **2017**, 1567, 379-390	1.4	4
142	CAD mutations and uridine-responsive epileptic encephalopathy. *Brain*, **2017**, 140, 279-286	11.2	77
141	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
140	Mutations in MDH2, Encoding a Krebs Cycle Enzyme, Cause Early-Onset Severe Encephalopathy. *American Journal of Human Genetics*, **2017**, 100, 151-159	11	38
139	Treatable mitochondrial diseases: cofactor metabolism and beyond. *Brain*, **2017**, 140, e11	11.2	46
138	Epigenome-wide association study of body mass index, and the adverse outcomes of adiposity. *Nature*, **2017**, 541, 81-86	50.4	511
137	CoQ deficiency causes disruption of mitochondrial sulfide oxidation, a new pathomechanism associated with this syndrome. *EMBO Molecular Medicine*, **2017**, 9, 78-95	12	47
136	Novel GFM2 variants associated with early-onset neurological presentations of mitochondrial disease and impaired expression of OXPHOS subunits. *Neurogenetics*, **2017**, 18, 227-235	3	5
135	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
134	High Symmetry of Visual Acuity and Visual Fields in RPGR-Linked Retinitis Pigmentosa **2017**, 58, 4457-4466		11
133	Biallelic variants in WARS2 encoding mitochondrial tryptophanyl-tRNA synthase in six individuals with mitochondrial encephalopathy. *Human Mutation*, **2017**, 38, 1786-1795	4.7	15
132	Molecular and clinical spectra of FBXL4 deficiency. *Human Mutation*, **2017**, 38, 1649-1659	4.7	29
131	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
130	Transcriptome-Wide Analysis Identifies Novel Associations With Blood Pressure. *Hypertension*, **2017**, 70, 743-750	8.5	21
129	LYRM7 - associated complex III deficiency: A clinical, molecular genetic, MR tomographic, and biochemical study. *Mitochondrion*, **2017**, 37, 55-61	4.9	17

128	Rare causes of early-onset dystonia-parkinsonism with cognitive impairment: a de novo PSEN-1 mutation. *Neurogenetics*, **2017**, 18, 175-178	3	13
127	ACSL4 dictates ferroptosis sensitivity by shaping cellular lipid composition. *Nature Chemical Biology* , **2017**, 13, 91-98	11.7	908
126	Neonatal encephalocardiomyopathy caused by mutations in VARS2. *Metabolic Brain Disease*, **2017**, 32, 267-270	3.9	21
125	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
124	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
123	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
122	A whole-blood transcriptome meta-analysis identifies gene expression signatures of cigarette smoking. *Human Molecular Genetics*, **2016**, 25, 4611-4623	5.6	58
121	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
120	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
119	Sudden Cardiac Death Due to Deficiency of the Mitochondrial Inorganic Pyrophosphatase PPA2. *American Journal of Human Genetics*, **2016**, 99, 674-682	11	33
118	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
117	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
116	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
115	Mutations in SLC39A14 disrupt manganese homeostasis and cause childhood-onset parkinsonism-dystonia. *Nature Communications*, **2016**, 7, 11601	17.4	160
114	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
113	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
112	Bezafibrate Improves Insulin Sensitivity and Metabolic Flexibility in STZ-Induced Diabetic Mice. *Diabetes*, **2016**, 65, 2540-52	0.9	28
111	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

110	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
109	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). *Autophagy*, **2016**, 12, 1-222	10.2	3838
108	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
107	EARS2 mutations cause fatal neonatal lactic acidosis, recurrent hypoglycemia and agenesis of corpus callosum. *Metabolic Brain Disease*, **2016**, 31, 717-21	3.9	12
106	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
105	Analyzing Illumina Gene Expression Microarray Data Obtained From Human Whole Blood Cell and Blood Monocyte Samples. *Methods in Molecular Biology*, **2016**, 1368, 85-97	1.4	2
104	Human thioredoxin 2 deficiency impairs mitochondrial redox homeostasis and causes early-onset neurodegeneration. *Brain*, **2016**, 139, 346-54	11.2	64
103	Atypical Clinical Presentations of TAZ Mutations: An Underdiagnosed Cause of Growth Retardation?. *JIMD Reports*, **2016**, 29, 89-93	1.9	7
102	Epigenetic Signatures at AQP3 and SOCS3 Engage in Low-Grade Inflammation across Different Tissues. *PLoS ONE*, **2016**, 11, e0166015	3.7	10
101	Association between DNA Methylation in Whole Blood and Measures of Glucose Metabolism: KORA F4 Study. *PLoS ONE*, **2016**, 11, e0152314	3.7	50
100	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
99	Mitochondrial replacement approaches: challenges for clinical implementation. *Genome Medicine*, **2016**, 8, 126	14.4	15
98	DNA methylation signatures of chronic low-grade inflammation are associated with complex diseases. *Genome Biology*, **2016**, 17, 255	18.3	171
97	Genetic cause and prevalence of hydroxyprolinemia. *Journal of Inherited Metabolic Disease*, **2016**, 39, 625-632	5.4	8
96	Severe respiratory complex III defect prevents liver adaptation to prolonged fasting. *Journal of Hepatology*, **2016**, 65, 377-85	13.4	23
95	Mitochondrial Protein Interaction Mapping Identifies Regulators of Respiratory Chain Function. *Molecular Cell*, **2016**, 63, 621-632	17.6	163
94	The many faces of paediatric mitochondrial disease on neuroimaging. *Childs Nervous System*, **2016**, 32, 2077-2083	1.7	14
93	DNA methylation of lipid-related genes affects blood lipid levels. *Circulation: Cardiovascular Genetics*, **2015**, 8, 334-42		122

92	Cell Specific eQTL Analysis without Sorting Cells. *PLoS Genetics*, **2015**, 11, e1005223	6	81
91	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
90	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
89	Biallelic Mutations in NBAS Cause Recurrent Acute Liver Failure with Onset in Infancy. *American Journal of Human Genetics*, **2015**, 97, 163-9	11	83
88	A meta-analysis of gene expression signatures of blood pressure and hypertension. *PLoS Genetics*, **2015**, 11, e1005035	6	83
87	MRPL44 mutations cause a slowly progressive multisystem disease with childhood-onset hypertrophic cardiomyopathy. *Neurogenetics*, **2015**, 16, 319-23	3	39
86	Spectrum of combined respiratory chain defects. *Journal of Inherited Metabolic Disease*, **2015**, 38, 629-40	5.4	78
85	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
84	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
83	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
82	The transcriptional landscape of age in human peripheral blood. *Nature Communications*, **2015**, 6, 8570	17.4	335
81	MRPS22 mutation causes fatal neonatal lactic acidosis with brain and heart abnormalities. *Neurogenetics*, **2015**, 16, 237-40	3	28
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