Thomas Meitinger

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/6625070/publications.pdf

Version: 2024-02-01

581	107,683	196	248
papers	citations	149 h-index	302 g-index

630	630	630	94995
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Biological, clinical and population relevance of 95 loci for blood lipids. Nature, 2010, 466, 707-713.	13.7	3,249
2	Mutations in LRRK2 Cause Autosomal-Dominant Parkinsonism with Pleomorphic Pathology. Neuron, 2004, 44, 601-607.	3.8	2,653
3	Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. Nature Genetics, 2010, 42, 937-948.	9.4	2,634
4	A reference panel of 64,976 haplotypes for genotype imputation. Nature Genetics, 2016, 48, 1279-1283.	9.4	2,421
5	A comprehensive 1000 Genomesâ€“based genome-wide association meta-analysis of coronary artery disease. Nature Genetics, 2015, 47, 1121-1130.	9.4	2,054
6	Genomewide Association Analysis of Coronary Artery Disease. New England Journal of Medicine, 2007, 357, 443-453.	13.9	1,865
7	Transcriptome and genome sequencing uncovers functional variation in humans. Nature, 2013, 501, 506-511.	13.7	1,857
8	Genetic variants in novel pathways influence blood pressure and cardiovascular disease risk. Nature, 2011, 478, 103-109.	13.7	1,855
9	Defining the role of common variation in the genomic and biological architecture of adult human height. Nature Genetics, 2014, 46, 1173-1186.	9.4	1,818
10	Hundreds of variants clustered in genomic loci and biological pathways affect human height. Nature, 2010, 467, 832-838.	13.7	1,789
11	Large-scale association analysis identifies 13 new susceptibility loci for coronary artery disease. Nature Genetics, 2011, 43, 333-338.	9.4	1,685
12	Twelve type 2 diabetes susceptibility loci identified through large-scale association analysis. Nature Genetics, 2010, 42, 579-589.	9.4	1,631
13	Six new loci associated with body mass index highlight a neuronal influence on body weight regulation. Nature Genetics, 2009, 41, 25-34.	9.4	1,572
14	Systematic identification of trans eQTLs as putative drivers of known disease associations. Nature Genetics, 2013, 45, 1238-1243.	9.4	1,544
15	Autosomal dominant hypophosphataemic rickets is associated with mutations in FGF23. Nature Genetics, 2000, 26, 345-348.	9.4	1,411
16	Fine-mapping type 2 diabetes loci to single-variant resolution using high-density imputation and islet-specific epigenome maps. Nature Genetics, 2018, 50, 1505-1513.	9.4	1,331
17	Genome-wide association study identifies 74 loci associated with educational attainment. Nature, 2016, 533, 539-542.	13.7	1,204
18	Common variants near MC4R are associated with fat mass, weight and risk of obesity. Nature Genetics, 2008, 40, 768-775.	9.4	1,179

#	Article	IF	Citations
19	Genome-wide association study identifies eight loci associated with blood pressure. Nature Genetics, 2009, 41, 666-676.	9.4	1,104
20	A gene (PEX) with homologies to endopeptidases is mutated in patients with Xâ€“linked hypophosphatemic rickets. Nature Genetics, 1995, 11, 130-136.	9.4	1,067
21	The DNA sequence of the human X chromosome. Nature, 2005, 434, 325-337.	13.7	985
22	The genetic architecture of type 2 diabetes. Nature, 2016, 536, 41-47.	13.7	952
23	Range of genetic mutations associated with severe non-syndromic sporadic intellectual disability: an exome sequencing study. Lancet, The, 2012, 380, 1674-1682.	6.3	940
24	Loss-of-Function Mutations inAPOC3,Triglycerides, and Coronary Disease. New England Journal of Medicine, 2014, 371, 22-31.	13.9	936
25	Human metabolic individuality in biomedical and pharmaceutical research. Nature, 2011, 477, 54-60.	13.7	916
26	Polymorphisms in FKBP5 are associated with increased recurrence of depressive episodes and rapid response to antidepressant treatment. Nature Genetics, 2004, 36, 1319-1325.	9.4	892
27	Meta-analysis identifies 13 new loci associated with waist-hip ratio and reveals sexual dimorphism in the genetic basis of fat distribution. Nature Genetics, 2010, 42, 949-960.	9.4	836
28	An unusual member of the nuclear hormone receptor superfamily responsible for X-linked adrenal hypoplasia congenita. Nature, 1994, 372, 635-641.	13.7	796
29	Loci influencing lipid levels and coronary heart disease risk in 16 European population cohorts. Nature Genetics, 2009, 41, 47-55.	9.4	776
30	A Mutation in VPS35, Encoding a Subunit of the Retromer Complex, Causes Late-Onset Parkinson Disease. American Journal of Human Genetics, 2011, 89, 168-175.	2.6	757
31	Epigenome-wide association study of body mass index, and the adverse outcomes of adiposity. Nature, 2017, 541, 81-86.	13.7	743
32	Hypothetical LOC387715 is a second major susceptibility gene for age-related macular degeneration, contributing independently of complement factor H to disease risk. Human Molecular Genetics, 2005, 14, 3227-3236.	1.4	741
33	Mutations in the DAX-1 gene give rise to both X-linked adrenal hypoplasia congenita and hypogonadotropic hypogonadism. Nature, 1994, 372, 672-676.	13.7	722
34	Seven new loci associated with age-related macular degeneration. Nature Genetics, 2013, 45, 433-439.	9.4	687
35	Genetics Meets Metabolomics: A Genome-Wide Association Study of Metabolite Profiles in Human Serum. PLoS Genetics, 2008, 4, e1000282.	1.5	660
36	Genome-wide, large-scale production of mutant mice by ENU mutagenesis. Nature Genetics, 2000, 25, 444-447.	9.4	658

#	Article	IF	Citations
37	A Common Genetic Variant Is Associated with Adult and Childhood Obesity. Science, 2006, 312, 279-283.	6.0	652
38	Haploinsufficiency of TBK1 causes familial ALS and fronto-temporal dementia. Nature Neuroscience, 2015, 18, 631-636.	7.1	652
39	Genome-wide association study of restless legs syndrome identifies common variants in three genomic regions. Nature Genetics, 2007, 39, 1000-1006.	9.4	633
40	A genome-wide perspective of genetic variation in human metabolism. Nature Genetics, 2010, 42, 137-141.	9.4	618
41	An Expanded Genome-Wide Association Study of Type 2 Diabetes in Europeans. Diabetes, 2017, 66, 2888-2902.	0.3	615
42	Mutations in the CEP290 (NPHP6) Gene Are a Frequent Cause of Leber Congenital Amaurosis. American Journal of Human Genetics, 2006, 79, 556-561.	2.6	608
43	Novel biomarkers for preâ€diabetes identified by metabolomics. Molecular Systems Biology, 2012, 8, 615.	3.2	605
44	Meta-Analysis of 28,141 Individuals Identifies Common Variants within Five New Loci That Influence Uric Acid Concentrations. PLoS Genetics, 2009, 5, e1000504.	1.5	572
45	A catalog of genetic loci associated with kidney function from analyses of a million individuals. Nature Genetics, 2019, 51, 957-972.	9.4	549
46	Meta-analysis identifies six new susceptibility loci for atrial fibrillation. Nature Genetics, 2012, 44, 670-675.	9.4	533
47	Mutations in the gene encoding É›-sarcoglycan cause myoclonusâ€“dystonia syndrome. Nature Genetics, 2001, 29, 66-69.	9.4	523
48	Meta-analysis of 375,000 individuals identifies 38 susceptibility loci for migraine. Nature Genetics, 2016, 48, 856-866.	9.4	520
49	Y-Chromosomal Diversity in Europe Is Clinal and Influenced Primarily by Geography, Rather than by Language. American Journal of Human Genetics, 2000, 67, 1526-1543.	2.6	519
50	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	3.8	517
51	Human TUBB3 Mutations Perturb Microtubule Dynamics, Kinesin Interactions, and Axon Guidance. Cell, 2010, 140, 74-87.	13.5	515
52	Mutations of SURF-1 in Leigh Disease Associated with Cytochrome c Oxidase Deficiency. American Journal of Human Genetics, 1998, 63, 1609-1621.	2.6	504
53	A common genetic variant in the NOS1 regulator NOS1AP modulates cardiac repolarization. Nature Genetics, 2006, 38, 644-651.	9.4	500
54	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1043-1048.	9.4	494

#	Article	IF	Citations
55	Somatic mutations in ATP1A1 and ATP2B3 lead to aldosterone-producing adenomas and secondary hypertension. Nature Genetics, 2013, 45, 440-444.	9.4	460
56	A gene (RPGR) with homology to the RCC1 guanine nucleotide exchange factor is mutated in Xâ€“linked retinitis pigmentosa (RP3). Nature Genetics, 1996, 13, 35-42.	9.4	453
57	Genome-Wide Association Scan Meta-Analysis Identifies Three Loci Influencing Adiposity and Fat Distribution. PLoS Genetics, 2009, 5, e1000508.	1.5	453
58	An L-type calcium-channel gene mutated in incomplete X-linked congenital stationary night blindness. Nature Genetics, 1998, 19, 260-263.	9.4	450
59	Mutations in the deubiquitinase gene USP8 cause Cushing's disease. Nature Genetics, 2015, 47, 31-38.	9.4	450
60	Narcolepsy is strongly associated with the T-cell receptor alpha locus. Nature Genetics, 2009, 41, 708-711.	9.4	445
61	The Parkinson disease causing LRRK2 mutation I2020T is associated with increased kinase activity. Human Molecular Genetics, 2006, 15, 223-232.	1.4	442
62	New susceptibility locus for coronary artery disease on chromosome 3q22.3. Nature Genetics, 2009, 41, 280-282.	9.4	440
63	Common variants in KCNN3 are associated with lone atrial fibrillation. Nature Genetics, 2010, 42, 240-244.	9.4	438
64	Genetic diagnosis of Mendelian disorders via RNA sequencing. Nature Communications, 2017, 8, 15824.	5.8	432
65	Loss-of-function mutations in SLC30A8 protect against type 2 diabetes. Nature Genetics, 2014, 46, 357-363.	9.4	428
66	Genome-wide haplotype association study identifies the SLC22A3-LPAL2-LPA gene cluster as a risk locus for coronary artery disease. Nature Genetics, 2009, 41, 283-285.	9.4	427
67	Coding Variation inANGPTL4,LPL,andSVEP1and the Risk of Coronary Disease. New England Journal of Medicine, 2016, 374, 1134-1144.	13.9	427
68	Genomic analyses identify hundreds of variants associated with age at menarche and support a role for puberty timing in cancer risk. Nature Genetics, 2017, 49, 834-841.	9.4	426
69	Novel Loci for Adiponectin Levels and Their Influence on Type 2 Diabetes and Metabolic Traits: A Multi-Ethnic Meta-Analysis of 45,891 Individuals. PLoS Genetics, 2012, 8, e1002607.	1.5	419
70	Diabetes insipidus, diabetes mellitus, optic atrophy and deafness (DIDMOAD) caused by mutations in a novel gene (wolframin) coding for a predicted transmembrane protein. Human Molecular Genetics, 1998, 7, 2021-2028.	1.4	403
71	Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. Nature Genetics, 2011, 43, 1005-1011.	9.4	403
72	Genome-wide association study of PR interval. Nature Genetics, 2010, 42, 153-159.	9.4	400

#	Article	IF	Citations
73	Genome-wide meta-analyses of multiancestry cohorts identify multiple new susceptibility loci for refractive error and myopia. Nature Genetics, 2013, 45, 314-318.	9.4	398
74	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. Nature, 2011, 478, 97-102.	13.7	394
75	Mutational hot spot within a new RPGR exon in X-linked retinitis pigmentosa. Nature Genetics, 2000, 25, 462-466.	9.4	392
76	Common Variants at 10 Genomic Loci Influence Hemoglobin A1C Levels via Glycemic and Nonglycemic Pathways. Diabetes, 2010, 59, 3229-3239.	0.3	387
77	Inactivating Mutations in NPC1L1 and Protection from Coronary Heart Disease. New England Journal of Medicine, 2014, 371, 2072-2082.	13.9	386
78	Genome-wide association study identifies two susceptibility loci for nonsyndromic cleft lip with or without cleft palate. Nature Genetics, 2010, 42, 24-26.	9.4	379
79	SLC2A9 influences uric acid concentrations with pronounced sex-specific effects. Nature Genetics, 2008, 40, 430-436.	9.4	363
80	Variants in ZFHX3 are associated with atrial fibrillation in individuals of European ancestry. Nature Genetics, 2009, 41, 879-881.	9.4	363
81	Common variants at ten loci modulate the QT interval duration in the QTSCD Study. Nature Genetics, 2009, 41, 407-414.	9.4	356
82	Refining the accuracy of validated target identification through coding variant fine-mapping in type 2 diabetes. Nature Genetics, 2018, 50, 559-571.	9.4	356
83	Constitutive Activation of PKA Catalytic Subunit in Adrenal Cushing's Syndrome. New England Journal of Medicine, 2014, 370, 1019-1028.	13.9	355
84	Impact of common genetic determinants of Hemoglobin A1c on type 2 diabetes risk and diagnosis in ancestrally diverse populations: A transethnic genome-wide meta-analysis. PLoS Medicine, 2017, 14, e1002383.	3.9	341
85	Genome-wide meta-analysis identifies new susceptibility loci for migraine. Nature Genetics, 2013, 45, 912-917.	9.4	338
86	Genome-wide association study of migraine implicates a common susceptibility variant on 8q22.1. Nature Genetics, 2010, 42, 869-873.	9.4	332
87	Calmodulin Mutations Associated With Recurrent Cardiac Arrest in Infants. Circulation, 2013, 127, 1009-1017.	1.6	331
88	Discovery of Sexual Dimorphisms in Metabolic and Genetic Biomarkers. PLoS Genetics, 2011, 7, e1002215.	1.5	328
89	Genome Analyses of >200,000 Individuals Identify 58 Loci for Chronic Inflammation and Highlight Pathways that Link Inflammation and Complex Disorders. American Journal of Human Genetics, 2018, 103, 691-706.	2.6	326
90	Mutations in a new gene in Ellis-van Creveld syndrome and Weyers acrodental dysostosis. Nature Genetics, 2000, 24, 283-286.	9.4	323

#	Article	IF	Citations
91	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
92	Common variants in 22 loci are associated with QRS duration and cardiac ventricular conduction. Nature Genetics, 2010, 42, 1068-1076.	9.4	308
93	Prevalence of refractive error in Europe: the European Eye Epidemiology (E3) Consortium. European Journal of Epidemiology, 2015, 30, 305-315.	2.5	306
94	Genome-wide association analysis identifies susceptibility loci for migraine without aura. Nature Genetics, 2012, 44, 777-782.	9.4	294
95	Trans-ancestry genome-wide association study identifies 12 genetic loci influencing blood pressure and implicates a role for DNA methylation. Nature Genetics, 2015, 47, 1282-1293.	9.4	294
96	The Autosomal Dominant Hypophosphatemic Rickets (ADHR) Gene Is a Secreted Polypeptide Overexpressed by Tumors that Cause Phosphate Wasting. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 497-500.	1.8	292
97	Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization. Nature Genetics, 2014, 46, 826-836.	9.4	281
98	Genetic Structure of Europeans: A View from the Northâ€“East. PLoS ONE, 2009, 4, e5472.	1.1	279
99	Characterization of circular RNAs in human, mouse and rat hearts. Journal of Molecular and Cellular Cardiology, 2016, 98, 103-107.	0.9	274
100	Genome-Wide Scan on Total Serum IgE Levels Identifies FCER1A as Novel Susceptibility Locus. PLoS Genetics, 2008, 4, e1000166.	1.5	255
101	PTPRD (protein tyrosine phosphatase receptor type delta) is associated with restless legs syndrome. Nature Genetics, 2008, 40, 946-948.	9.4	252
102	Effect of predicted protein-truncating genetic variants on the human transcriptome. Science, 2015, 348, 666-669.	6.0	252
103	Target genes, variants, tissues and transcriptional pathways influencing human serum urate levels. Nature Genetics, 2019, 51, 1459-1474.	9.4	251
104	Multi-ancestry genetic study of type 2 diabetes highlights the power of diverse populations for discovery and translation. Nature Genetics, 2022, 54, 560-572.	9.4	250
105	Exome sequencing of 20,791Âcases of type 2 diabetes and 24,440Âcontrols. Nature, 2019, 570, 71-76.	13.7	248
106	A powerful tool for genome analysis in maize: development and evaluation of the high density 600 k SNP genotyping array. BMC Genomics, 2014, 15, 823.	1.2	242
107	Isolation of a candidate gene for Norrie disease by positional cloning. Nature Genetics, 1992, 1, 199-203.	9.4	239
108	Genome-wide association meta-analysis highlights light-induced signaling as a driver for refractive error. Nature Genetics, 2018, 50, 834-848.	9.4	239

#	Article	IF	Citations
109	Exome sequencing identifies ACAD9 mutations as a cause of complex I deficiency. Nature Genetics, 2010, 42, 1131-1134.	9.4	234
110	Dysfunctional nitric oxide signalling increases risk of myocardial infarction. Nature, 2013, 504, 432-436.	13.7	230
111	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
112	Multiple regions of Î±-synuclein are associated with Parkinson's disease. Annals of Neurology, 2005, 57, 535-541.	2.8	223
113	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042.	9.4	218
114	Common variants in P2RY11 are associated with narcolepsy. Nature Genetics, 2011, 43, 66-71.	9.4	215
115	Polygenic prediction of educational attainment within and between families from genome-wide association analyses in 3 million individuals. Nature Genetics, 2022, 54, 437-449.	9.4	215
116	A common region of 10p deleted in DiGeorge and velocardiofacial syndromes. Nature Genetics, 1996, 13, 458-460.	9.4	214
117	Systematic Evaluation of Pleiotropy Identifies 6 Further Loci Associated WithÂCoronary ArteryÂDisease. Journal of the American College of Cardiology, 2017, 69, 823-836.	1.2	214
118	Genetic Adaptation of Fatty-Acid Metabolism: A Human-Specific Haplotype Increasing the Biosynthesis of Long-Chain Omega-3 and Omega-6 Fatty Acids. American Journal of Human Genetics, 2012, 90, 809-820.	2.6	205
119	Beta-propeller protein-associated neurodegeneration: a new X-linked dominant disorder with brain iron accumulation. Brain, 2013, 136, 1708-1717.	3.7	203
120	Genetic Variants Associated With Cardiac Structure and Function. JAMA - Journal of the American Medical Association, 2009, 302, 168.	3.8	202
121	Sequence variants on chromosome 9p21.3 confer risk for atherosclerotic stroke. Annals of Neurology, 2009, 65, 531-539.	2.8	199
122	Loss-of-function mutations in MGME1 impair mtDNA replication and cause multisystemic mitochondrial disease. Nature Genetics, 2013, 45, 214-219.	9.4	198
123	A Genome-Wide Association Search for Type 2 Diabetes Genes in African Americans. PLoS ONE, 2012, 7, e29202.	1.1	197
124	Pex Gene Deletions in Gy and Hyp Mice Provide Mouse Models for X-Linked Hypophosphatemia. Human Molecular Genetics, 1997, 6, 165-171.	1.4	195
125	Large scale replication and meta-analysis of variants on chromosome 4q25 associated with atrial fibrillation. European Heart Journal, 2008, 30, 813-819.	1.0	193
126	Whole-Exome Sequencing Identifies Rare and Low-Frequency Coding Variants Associated with LDL Cholesterol. American Journal of Human Genetics, 2014, 94, 233-245.	2.6	193

#	Article	IF	Citations
127	Lack of the Mitochondrial Protein Acylglycerol Kinase Causes Sengers Syndrome. American Journal of Human Genetics, 2012, 90, 314-320.	2.6	192
128	Novel loci affecting iron homeostasis and their effects in individuals at risk for hemochromatosis. Nature Communications, 2014, 5, 4926.	5.8	192
129	Intragenic Deletion of theKALIG-1Gene in Kallmann's Syndrome. New England Journal of Medicine, 1992, 326, 1752-1755.	13.9	189
130	Genome-Wide Association Studies of Serum Magnesium, Potassium, and Sodium Concentrations Identify Six Loci Influencing Serum Magnesium Levels. PLoS Genetics, 2010, 6, e1001045.	1.5	185
131	Distribution of mutations in the PEX gene in families with X-linked hypophosphataemic rickets (HYP). Human Molecular Genetics, 1997, 6, 539-549.	1.4	184
132	Genotypic and phenotypic spectrum of PANK2 mutations in patients with neurodegeneration with brain iron accumulation. Annals of Neurology, 2006, 59, 248-256.	2.8	184
133	Genetic Determinants of Circulating Sphingolipid Concentrations in European Populations. PLoS Genetics, 2009, 5, e1000672.	1.5	184
134	A genome-wide association study confirms APOE as the major gene influencing survival in long-lived individuals. Mechanisms of Ageing and Development, 2011, 132, 324-330.	2.2	184
135	Genetic insights into biological mechanisms governing human ovarian ageing. Nature, 2021, 596, 393-397.	13.7	183
136	Molecular diagnosis in mitochondrial complex I deficiency using exome sequencing. Journal of Medical Genetics, 2012, 49, 277-283.	1.5	182
137	Integrative Analysis of the Mitochondrial Proteome in Yeast. PLoS Biology, 2004, 2, e160.	2.6	181
138	Prevalence, Spectrum, and Functional Characterization of Melanocortin-4 Receptor Gene Mutations in a Representative Population-Based Sample and Obese Adults from Germany. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 1761-1769.	1.8	181
139	Genome-Wide Association Study Identifies Novel Loci Associated with Circulating Phospho- and Sphingolipid Concentrations. PLoS Genetics, 2012, 8, e1002490.	1.5	181
140	Genetic Determinants of Serum Testosterone Concentrations in Men. PLoS Genetics, 2011, 7, e1002313.	1.5	178
141	Chronic recurrent multifocal osteomyelitis (CRMO): evidence for a susceptibility gene located on chromosome 18q21.3-18q22. European Journal of Human Genetics, 2002, 10, 217-221.	1.4	176
142	ALOX5AP Gene and the PDE4D Gene in a Central European Population of Stroke Patients. Stroke, 2005, 36, 731-736.	1.0	174
143	Directional dominance on stature and cognition inÂdiverse human populations. Nature, 2015, 523, 459-462.	13.7	173
144	Novel (ovario) leukodystrophy related to AARS2 mutations. Neurology, 2014, 82, 2063-2071.	1.5	172

#	Article	IF	Citations
145	Genome-wide scan identifies CDH13 as a novel susceptibility locus contributing to blood pressure determination in two European populations. Human Molecular Genetics, 2009, 18, 2288-2296.	1.4	170
146	Genome-wide association study identifies a susceptibility locus at 21q21 for ventricular fibrillation in acute myocardial infarction. Nature Genetics, 2010, 42, 688-691.	9.4	170
147	Association of genetic variation with systolic and diastolic blood pressure among African Americans: the Candidate Gene Association Resource study. Human Molecular Genetics, 2011, 20, 2273-2284.	1.4	168
148	Hot-spot KIF5A mutations cause familial ALS. Brain, 2018, 141, 688-697.	3.7	167
149	Genome-wide association study of survival from sepsis due to pneumonia: an observational cohort study. Lancet Respiratory Medicine,the, 2015, 3, 53-60.	5.2	166
150	Molecular modelling of the Norrie disease protein predicts a cystine knot growth factor tertiary structure. Nature Genetics, 1993, 5, 376-380.	9.4	165
151	Novel Calmodulin Mutations Associated With Congenital Arrhythmia Susceptibility. Circulation: Cardiovascular Genetics, 2014, 7, 466-474.	5.1	165
152	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
153	Genome-Wide Association Study Identifies Novel Restless Legs Syndrome Susceptibility Loci on 2p14 and 16q12.1. PLoS Genetics, 2011, 7, e1002171.	1.5	163
154	Genomic Organization of the Human PEX Gene Mutated in X-Linked Dominant Hypophosphatemicâ€‰Rickets. Genome Research, 1997, 7, 573-585.	2.4	159
155	Shared genetic pathways contribute to risk of hypertrophic and dilated cardiomyopathies with opposite directions of effect. Nature Genetics, 2021, 53, 128-134.	9.4	155
156	Rare variants in PPARG with decreased activity in adipocyte differentiation are associated with increased risk of type 2 diabetes. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 13127-13132.	3.3	152
157	A Large Candidate Gene Survey Identifies the KCNE1 D85N Polymorphism as a Possible Modulator of Drug-Induced Torsades de Pointes. Circulation: Cardiovascular Genetics, 2012, 5, 91-99.	5.1	150
158	Improved proteome analysis of Saccharomyces cerevisiae mitochondria by free-flow electrophoresis. Proteomics, 2003, 3, 906-916.	1.3	148
159	The epsilon-sarcoglycan gene (SGCE), mutated in myoclonus-dystonia syndrome, is maternally imprinted. European Journal of Human Genetics, 2003, 11, 138-144.	1.4	148
160	Large meta-analysis of genome-wide association studies identifies five loci for lean body mass. Nature Communications, 2017, 8, 80.	5.8	147
161	Association of the PHACTR1/EDN1 Genetic Locus With Spontaneous Coronary Artery Dissection. Journal of the American College of Cardiology, 2019, 73, 58-66.	1.2	147
162	Norrie disease is caused by mutations in an extracellular protein resembling Câ€“terminal globular domain of mucins. Nature Genetics, 1992, 2, 139-143.	9.4	145

#	Article	IF	Citations
163	Meta-Analysis of Genome-Wide Association Studies Identifies Six New Loci for Serum Calcium Concentrations. PLoS Genetics, 2013, 9, e1003796.	1.5	142
164	Genetic Regulation of Serum Phytosterol Levels and Risk of Coronary Artery Disease. Circulation: Cardiovascular Genetics, 2010, 3, 331-339.	5.1	141
165	Genetic variation at chromosome 1p13.3 affects sortilin mRNA expression, cellular LDL-uptake and serum LDL levels which translates to the risk of coronary artery disease. Atherosclerosis, 2010, 208, 183-189.	0.4	141
166	Nine Loci for Ocular Axial Length Identified through Genome-wide Association Studies, Including Shared Loci with Refractive Error. American Journal of Human Genetics, 2013, 93, 264-277.	2.6	139
167	Common Variants in Myocardial Ion Channel Genes Modify the QT Interval in the General Population. Circulation Research, 2005, 96, 693-701.	2.0	138
168	Mutations in FBXL4, Encoding a Mitochondrial Protein, Cause Early-Onset Mitochondrial Encephalomyopathy. American Journal of Human Genetics, 2013, 93, 482-495.	2.6	138
169	Lifelong Reduction of LDL-Cholesterol Related to a Common Variant in the LDL-Receptor Gene Decreases the Risk of Coronary Artery Diseaseâ€"A Mendelian Randomisation Study. PLoS ONE, 2008, 3, e2986.	1.1	137
170	A genome-wide association study identifies 6p21 as novel risk locus for dilated cardiomyopathy. European Heart Journal, 2014, 35, 1069-1077.	1.0	137
171	The Association of a SNP Upstream of INSIG2 with Body Mass Index is Reproduced in Several but Not All Cohorts. PLoS Genetics, 2007, 3, e61.	1.5	134
172	Mutation Update for Kabuki Syndrome GenesKMT2DandKDM6Aand Further Delineation of X-Linked Kabuki Syndrome Subtype 2. Human Mutation, 2016, 37, 847-864.	1.1	134
173	Genome-wide association analysis identifies multiple loci related to resting heart rate. Human Molecular Genetics, 2010, 19, 3885-3894.	1.4	133
174	Novel multiple sclerosis susceptibility loci implicated in epigenetic regulation. Science Advances, 2016, 2, e1501678.	4.7	133
175	Genome-wide association meta-analyses and fine-mapping elucidate pathways influencing albuminuria. Nature Communications, 2019, 10, 4130.	5.8	133
176	A KATP channel gene effect on sleep duration: from genome-wide association studies to function in Drosophila. Molecular Psychiatry, 2013, 18, 122-132.	4.1	132
177	CLOCK Gene Variants Associate with Sleep Duration in Two Independent Populations. Biological Psychiatry, 2010, 67, 1040-1047.	0.7	128
178	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
179	ELAC2 Mutations Cause a Mitochondrial RNA Processing Defect Associated with Hypertrophic Cardiomyopathy. American Journal of Human Genetics, 2013, 93, 211-223.	2.6	127
180	Dense genotyping of immune-related loci in idiopathic inflammatory myopathies confirms HLA alleles as the strongest genetic risk factor and suggests different genetic background for major clinical subgroups. Annals of the Rheumatic Diseases, 2016, 75, 1558-1566.	0.5	127

#	Article	IF	Citations
181	Biallelic Mutations in DNAJC12 Cause Hyperphenylalaninemia, Dystonia, and Intellectual Disability. American Journal of Human Genetics, 2017, 100, 257-266.	2.6	127
182	Mutations in the candidate gene for Norrie disease. Human Molecular Genetics, 1992, 1, 461-465.	1.4	126
183	Ghrelin Receptor Gene: Identification of Several Sequence Variants in Extremely Obese Children and Adolescents, Healthy Normal-Weight and Underweight Students, and Children with Short Normal Stature. Journal of Clinical Endocrinology and Metabolism, 2004, 89, 157-162.	1.8	126
184	Genome-wide association study identifies a new locus for coronary artery disease on chromosome 10p11.23. European Heart Journal, 2011, 32, 158-168.	1.0	124
185	Common Genetic Variants Associate with Serum Phosphorus Concentration. Journal of the American Society of Nephrology: JASN, 2010, 21, 1223-1232.	3.0	123
186	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
187	A Large-Scale Multi-ancestry Genome-wide Study Accounting for Smoking Behavior Identifies Multiple Significant Loci for Blood Pressure. American Journal of Human Genetics, 2018, 102, 375-400.	2.6	123
188	SNP-Based Analysis of Genetic Substructure in the German Population. Human Heredity, 2006, 62, 20-29.	0.4	121
189	Genome-wide methylation data mirror ancestry information. Epigenetics and Chromatin, 2017, 10, 1.	1.8	120
190	The retinitis pigmentosa GTPase regulator, RPGR, interacts with the delta subunit of rod cyclic GMP phosphodiesterase. Proceedings of the National Academy of Sciences of the United States of America, 1999, 96, 1315-1320.	3.3	119
191	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
192	Linkage Disequilibrium Patterns and tagSNP Transferability among European Populations. American Journal of Human Genetics, 2005, 76, 387-398.	2.6	117
193	An isoform of hPANK2, deficient in pantothenate kinase-associated neurodegeneration, localizes to mitochondria. Human Molecular Genetics, 2003, 12, 321-327.	1.4	115
194	Cell Specific eQTL Analysis without Sorting Cells. PLoS Genetics, 2015, 11, e1005223.	1.5	115
195	Copy-Number Variations Measured by Single-Nucleotideâ€“Polymorphism Oligonucleotide Arrays in Patients with Mental Retardation. American Journal of Human Genetics, 2007, 81, 768-779.	2.6	114
196	A genome-wide association meta-analysis on lipoprotein (a) concentrations adjusted for apolipoprotein (a) isoforms. Journal of Lipid Research, 2017, 58, 1834-1844.	2.0	114
197	Leveraging Cross-Species Transcription Factor Binding Site Patterns: From Diabetes Risk Loci to Disease Mechanisms. Cell, 2014, 156, 343-358.	13.5	113
198	52 Genetic Loci Influencing MyocardialÂMass. Journal of the American College of Cardiology, 2016, 68, 1435-1448.	1.2	113

#	Article	IF	Citations
199	A new family of interspersed repetitive DNA sequences in the mouse genome. Journal of Molecular Biology, 1982, 157, 453-471.	2.0	112
200	Multi-ancestry genome-wide geneâ€“smoking interaction study of 387,272 individuals identifies new loci associated with serum lipids. Nature Genetics, 2019, 51, 636-648.	9.4	112
201	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
202	Genetic Association Study Identifies HSPB7 as a Risk Gene for Idiopathic Dilated Cardiomyopathy. PLoS Genetics, 2010, 6, e1001167.	1.5	110
203	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
204	Genetic variants linked to education predict longevity. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 13366-13371.	3.3	110
205	A novel deletion in progranulin gene is associated with FTDP-17 and CBS. Neurobiology of Aging, 2008, 29, 427-435.	1.5	109
206	A Meta-analysis of Gene Expression Signatures of Blood Pressure and Hypertension. PLoS Genetics, 2015, 11, e1005035.	1.5	107
207	CADmutations and uridine-responsive epileptic encephalopathy. Brain, 2017, 140, 279-286.	3.7	106
208	NEK1mutations in familial amyotrophic lateral sclerosis. Brain, 2016, 139, e28-e28.	3.7	105
209	Variation in the 4q25 Chromosomal Locus Predicts Atrial Fibrillation After Coronary Artery Bypass Graft Surgery. Circulation: Cardiovascular Genetics, 2009, 2, 499-506.	5.1	104
210	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
211	Meta-analysis of geneâ€“environment-wide association scans accounting for education level identifies additional loci for refractive error. Nature Communications, 2016, 7, 11008.	5.8	104
212	X-linked Retinitis Pigmentosa:RPGRMutations in Most Families with Definite X Linkage and Clustering of Mutations in a Short Sequence Stretch of Exon ORF15. , 2003, 44, 1458.		103
213	Variants in the neuronal nitric oxide synthase (nNOS, NOS1) gene are associated with restless legs syndrome. Movement Disorders, 2008, 23, 350-358.	2.2	103
214	The non-synonymous coding IKr-channel variant KCNH2-K897T is associated with atrial fibrillation: results from a systematic candidate gene-based analysis of KCNH2 (HERG). European Heart Journal, 2008, 29, 907-914.	1.0	103
215	Restless Legs Syndrome-associated intronic common variant in Meis1 alters enhancer function in the developing telencephalon. Genome Research, 2014, 24, 592-603.	2.4	102
216	Spectrum of combined respiratory chain defects. Journal of Inherited Metabolic Disease, 2015, 38, 629-640.	1.7	102

#	Article	IF	Citations
217	Two novel mutations in conserved codons indicate that CHCHD10 is a gene associated with motor neuron disease. Brain, 2014, 137, e309-e309.	3.7	101
218	Absence of BiP Co-chaperone DNAJC3 Causes Diabetes Mellitus and Multisystemic Neurodegeneration. American Journal of Human Genetics, 2014, 95, 689-697.	2.6	100
219	Periodontal Ehlers-Danlos Syndrome Is Caused by Mutations in C1R and C1S , which Encode Subcomponents C1r and C1s of Complement. American Journal of Human Genetics, 2016, 99, 1005-1014.	2.6	100
220	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
221	Targeting 160 Candidate Genes for Blood Pressure Regulation with a Genome-Wide Genotyping Array. PLoS ONE, 2009, 4, e6034.	1.1	98
222	1000 Genomes-based meta-analysis identifies 10 novel loci for kidney function. Scientific Reports, 2017, 7, 45040.	1.6	98
223	Effects of Metformin on Metabolite Profiles and LDL Cholesterol in Patients With Type 2 Diabetes. Diabetes Care, 2015, 38, 1858-1867.	4.3	97
224	DHTKD1 Mutations Cause 2-Aminoadipic and 2-Oxoadipic Aciduria. American Journal of Human Genetics, 2012, 91, 1082-1087.	2.6	94
225	Biallelic Mutations of Methionyl-tRNA Synthetase Cause a Specific Type of Pulmonary Alveolar Proteinosis Prevalent on RÃ©union Island. American Journal of Human Genetics, 2015, 96, 826-831.	2.6	94
226	Novel genetic associations for blood pressure identified via gene-alcohol interaction in up to 570K individuals across multiple ancestries. PLoS ONE, 2018, 13, e0198166.	1.1	94
227	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
228	Identification of Novel Mutations in Patients with Leber Congenital Amaurosis and Juvenile RP by Genome-wide Homozygosity Mapping with SNP Microarrays. , 2007, 48, 5690.		90
229	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
230	Common variants in the JAZF1 gene associated with height identified by linkage and genome-wide association analysis. Human Molecular Genetics, 2009, 18, 373-380.	1.4	88
231	Whole genome sequencing of a single Bos taurus animal for single nucleotide polymorphism discovery. Genome Biology, 2009, 10, R82.	13.9	88
232	Genome-Wide Association Analysis of High-Density Lipoprotein Cholesterol in the Population-Based KORA Study Sheds New Light on Intergenic Regions. Circulation: Cardiovascular Genetics, 2008, 1, 10-20.	5.1	87
233	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
234	VARS2andTARS2Mutations in Patients with Mitochondrial Encephalomyopathies. Human Mutation, 2014, 35, 983-989.	1.1	86

#	Article	IF	Citations
235	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
236	Human thioredoxin 2 deficiency impairs mitochondrial redox homeostasis and causes early-onset neurodegeneration. Brain, 2016, 139, 346-354.	3.7	86
237	Multiancestry Genome-Wide Association Study of Lipid Levels Incorporating Gene-Alcohol Interactions. American Journal of Epidemiology, 2019, 188, 1033-1054.	1.6	85
238	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38.	3.6	85
239	Mutations in the mitochondrial thioredoxin reductase gene TXNRD2 cause dilated cardiomyopathy. European Heart Journal, 2011, 32, 1121-1133.	1.0	84
240	Associations of autozygosity with a broad range of human phenotypes. Nature Communications, 2019, 10, 4957.	5.8	84
241	Comprehensive catalog of European biobanks. Nature Biotechnology, 2011, 29, 795-797.	9.4	83
242	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
243	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
244	Transethnic Genome-Wide Association Study Provides Insights in the Genetic Architecture and Heritability of Long QT Syndrome. Circulation, 2020, 142, 324-338.	1.6	83
245	Mutations in ATP6V1E1 or ATP6V1A Cause Autosomal-Recessive Cutis Laxa. American Journal of Human Genetics, 2017, 100, 216-227.	2.6	82
246	Deletion mapping on chromosome 10p and definition of a critical region for the second DiGeorge syndrome locus (DGS2). European Journal of Human Genetics, 1998, 6, 213-225.	1.4	81
247	Clinical, biochemical, and genetic spectrum of seven patients with NFU1 deficiency. Frontiers in Genetics, 2015, 06, 123.	1.1	81
248	Comprehensive analysis of the mutation spectrum in 301 German ALS families. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 817-827.	0.9	80
249	Evidence for further genetic locus heterogeneity and confirmation of RLS-1 in restless legs syndrome. Movement Disorders, 2006, 21, 28-33.	2.2	79
250	Chapter 1 The Mitochondrial Proteome Database. Methods in Enzymology, 2009, 457, 3-20.	0.4	78
251	Replication of restless legs syndrome loci in three European populations. Journal of Medical Genetics, 2009, 46, 315-318.	1.5	78
252	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

#	Article	IF	Citations
253	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
254	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
255	Molecular cloning of a novel human UDP-GalNAc:polypeptide N-acetylgalactosaminyltransferase, GalNAc-T8, and analysis as a candidate autosomal dominant hypophosphatemic rickets (ADHR) gene. Gene, 2000, 246, 347-356.	1.0	74
256	Proteome analysis of mitochondrial outer membrane fromNeurospora crassa. Proteomics, 2006, 6, 72-80.	1.3	74
257	Effects of Long-Term Averaging of Quantitative Blood Pressure Traits on the Detection of Genetic Associations. American Journal of Human Genetics, 2014, 95, 49-65.	2.6	73
258	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
259	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
260	Genetic Variation in Soluble Epoxide Hydrolase (EPHX2) Is Associated With an Increased Risk of Ischemic Stroke in White Europeans. Stroke, 2008, 39, 1593-1596.	1.0	71
261	Genome-wide linkage analysis of serum creatinine in three isolated European populations. Kidney International, 2009, 76, 297-306.	2.6	71
262	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
263	PR interval genome-wide association meta-analysis identifies 50 loci associated with atrial and atrioventricular electrical activity. Nature Communications, 2018, 9, 2904.	5.8	71
264	Systematic analysis of variants related to familial hypercholesterolemia in families with premature myocardial infarction. European Journal of Human Genetics, 2016, 24, 191-197.	1.4	70
265	MitoP2: An Integrative Tool for the Analysis of the Mitochondrial Proteome. Molecular Biotechnology, 2008, 40, 306-315.	1.3	69
266	RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome. Journal of Clinical Investigation, 2015, 125, 3585-3599.	3.9	69
267	A high risk phenotype of hypertrophic cardiomyopathy associated with a compound genotype of two mutated Î²-myosin heavy chain genes. Human Genetics, 1998, 102, 299-304.	1.8	68
268	Presence of a Major WFS1 Mutation in Spanish Wolfram Syndrome Pedigrees. Molecular Genetics and Metabolism, 2001, 72, 72-81.	0.5	67
269	A Genome-Wide Association Scan of RR and QT Interval Duration in 3 European Genetically Isolated Populations. Circulation: Cardiovascular Genetics, 2009, 2, 322-328.	5.1	67
270	Large scale international replication and meta-analysis study confirms association of the 15q14 locus with myopia. The CREAM consortium. Human Genetics, 2012, 131, 1467-1480.	1.8	67

#	Article	IF	Citations
271	Network inference from glycoproteomics data reveals new reactions in the IgG glycosylation pathway. Nature Communications, 2017, 8, 1483.	5.8	67
272	Genome-wide analysis of PDX1 target genes in human pancreatic progenitors. Molecular Metabolism, 2018, 9, 57-68.	3.0	67
273	MicroRNA-21â€“Dependent Macrophage-to-Fibroblast Signaling Determines the Cardiac Response to Pressure Overload. Circulation, 2021, 143, 1513-1525.	1.6	67
274	The Eye-of-the-Tiger Sign is not a Reliable Disease Marker for Hallervorden-Spatz Syndrome. Neuropediatrics, 2005, 36, 221-222.	0.3	66
275	Genome-wide association studies of atrial fibrillation: past, present, and future. Cardiovascular Research, 2011, 89, 701-709.	1.8	66
276	A One Base Pair Deletion in the Canine ATP13A2 Gene Causes Exon Skipping and Late-Onset Neuronal Ceroid Lipofuscinosis in the Tibetan Terrier. PLoS Genetics, 2011, 7, e1002304.	1.5	66
277	Interplay of cellâ€“cell contacts and RhoA/ <scp>MRTF</scp> â€A signaling regulates cardiomyocyte identity. EMBO Journal, 2018, 37, .	3.5	66
278	Genome-Wide Association Study on Immunoglobulin G Glycosylation Patterns. Frontiers in Immunology, 2018, 9, 277.	2.2	66
279	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
280	Analysis of the human GDNF gene reveals an inducible promoter, three exons, a triplet repeat within the 3'-UTR and alternative splice products. Human Molecular Genetics, 1998, 7, 1873-1886.	1.4	64
281	Assessment of the genomic variation in a cattle population by re-sequencing of key animals at low to medium coverage. BMC Genomics, 2013, 14, 446.	1.2	64
282	Genetic characterization of northeastern Italian population isolates in the context of broader European genetic diversity. European Journal of Human Genetics, 2013, 21, 659-665.	1.4	64
283	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
284	CNV-association meta-analysis in 191,161 European adults reveals new loci associated with anthropometric traits. Nature Communications, 2017, 8, 744.	5.8	64
285	Multi-ancestry study of blood lipid levels identifies four loci interacting with physical activity. Nature Communications, 2019, 10, 376.	5.8	64
286	De novo variants in neurodevelopmental disordersâ€”experiences from a tertiary care center. Clinical Genetics, 2021, 100, 14-28.	1.0	64
287	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
288	Novel association to the proprotein convertase PCSK7 gene locus revealed by analysing soluble transferrin receptor (sTfR) levels. Human Molecular Genetics, 2011, 20, 1042-1047.	1.4	62

#	Article	IF	Citations
289	Multi-ancestry sleep-by-SNP interaction analysis in 126,926 individuals reveals lipid loci stratified by sleep duration. Nature Communications, 2019, 10, 5121.	5.8	62
290	Suggestive evidence for linkage for restless legs syndrome on chromosome 19p13. Neurogenetics, 2008, 9, 75-82.	0.7	61
291	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
292	Meta-analysis of genome-wide association studies in five cohorts reveals common variants in RBFOX1, a regulator of tissue-specific splicing, associated with refractive error. Human Molecular Genetics, 2013, 22, 2754-2764.	1.4	60
293	Genetic variation influencing DNA methylation provides insights into molecular mechanisms regulating genomic function. Nature Genetics, 2022, 54, 18-29.	9.4	60
294	MEIS1 and BTBD9: genetic association with restless leg syndrome in end stage renal disease. Journal of Medical Genetics, 2011, 48, 462-466.	1.5	59
295	A comprehensive evaluation of the genetic architecture of sudden cardiac arrest. European Heart Journal, 2018, 39, 3961-3969.	1.0	59
296	Multi-ancestry GWAS of the electrocardiographic PR interval identifies 202 loci underlying cardiac conduction. Nature Communications, 2020, 11, 2542.	5.8	59
297	DNA, FISH and complementation studies in ICF syndrome: DNA hypomethylation of repetitive and single copy loci and evidence for a trans acting factor. Human Genetics, 1995, 96, 562-571.	1.8	58
298	Spermine deficiency in Gy mice caused by deletion of the spermine synthase gene. Human Molecular Genetics, 1998, 7, 541-547.	1.4	58
299	A meta-analysis of genome-wide association studies of the electrocardiographic early repolarization pattern. Heart Rhythm, 2012, 9, 1627-1634.	0.3	58
300	Macrocytic Anemia and Mitochondriopathy Resulting from a Defect in Sideroflexin 4. American Journal of Human Genetics, 2013, 93, 906-914.	2.6	58
301	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
302	Point mutations in the PDX1 transactivation domain impair human Î²-cell development and function. Molecular Metabolism, 2019, 24, 80-97.	3.0	58
303	Risk gene variants for nicotine dependence in the CHRNA5â€“CHRNA3â€“CHRNB4 cluster are associated with cognitive performance. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 1448-1458.	1.1	57
304	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
305	Epigenetics meets proteomics in an epigenome-wide association study with circulating blood plasma protein traits. Nature Communications, 2020, 11, 15.	5.8	57
306	The genetic study of three population microisolates in South Tyrol (MICROS): study design and epidemiological perspectives. BMC Medical Genetics, 2007, 8, 29.	2.1	56

#	Article	IF	Citations
307	Genetic Variation in Members of the Leukotriene Biosynthesis Pathway Confer an Increased Risk of Ischemic Stroke. Stroke, 2008, 39, 1109-1114.	1.0	55
308	Compound Heterozygosity of Low-Frequency Promoter Deletions and Rare Loss-of-Function Mutations in TXNL4A Causes Burn-McKeown Syndrome. American Journal of Human Genetics, 2014, 95, 698-707.	2.6	55
309	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
310	Predicting cardiac electrical response to sodium-channel blockade and Brugada syndrome using polygenic risk scores. European Heart Journal, 2019, 40, 3097-3107.	1.0	55
311	Genome-wide association analyses identify new Brugada syndrome risk loci and highlight a new mechanism of sodium channel regulation in disease susceptibility. Nature Genetics, 2022, 54, 232-239.	9.4	55
312	A gene (SRPX) encoding a sushi-repeat-containing protein is deleted in patients with X-linked retinitis pigmentosa. Human Molecular Genetics, 1995, 4, 2339-2346.	1.4	54
313	Missense mutations in the NDP gene in patients with a less severe course of Norrie disease. Human Molecular Genetics, 1995, 4, 489-490.	1.4	54
314	Familial Mental Retardation Syndrome ATR-16 Due to an Inherited Cryptic Subtelomeric Translocation, t(3;16)(q29;p13.3). American Journal of Human Genetics, 2000, 66, 16-25.	2.6	54
315	Identification of recurring tumor-specific somatic mutations in acute myeloid leukemia by transcriptome sequencing. Leukemia, 2011, 25, 821-827.	3.3	54
316	DeepWAS: Multivariate genotype-phenotype associations by directly integrating regulatory information using deep learning. PLoS Computational Biology, 2020, 16, e1007616.	1.5	54
317	Common variants of LRRK2 are not associated with sporadic Parkinson's disease. Annals of Neurology, 2005, 58, 905-908.	2.8	53
318	Exome sequencing revealed a splice site variant in the IQCE gene underlying post-axial polydactyly type A restricted to lower limb. European Journal of Human Genetics, 2017, 25, 960-965.	1.4	53
319	Targeted Resequencing and Systematic InÂVivo Functional Testing Identifies Rare Variants in MEIS1 as Significant Contributors to Restless Legs Syndrome. American Journal of Human Genetics, 2014, 95, 85-95.	2.6	52
320	Electroretinography as a Screening Method for Mutations Causing Retinal Dysfunction in Mice. , 2004, 45, 601.		51
321	Modeling of Environmental Effects in Genome-Wide Association Studies Identifies SLC2A2 and HP as Novel Loci Influencing Serum Cholesterol Levels. PLoS Genetics, 2010, 6, e1000798.	1.5	51
322	Analysis for Genetic Modifiers of Disease Severity in Patients With Long-QT Syndrome Type 2. Circulation: Cardiovascular Genetics, 2015, 8, 447-456.	5.1	51
323	Rare variants in Î²-Amyloid precursor protein (APP) and Parkinsonâ€™s disease. European Journal of Human Genetics, 2015, 23, 1328-1333.	1.4	50
324	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

#	Article	IF	Citations
325	Genome-wide association study identifies multiple new loci associated with Ewing sarcoma susceptibility. Nature Communications, 2018, 9, 3184.	5.8	50
326	Niemann-Pick C Disease Gene Mutations and Age-Related Neurodegenerative Disorders. PLoS ONE, 2013, 8, e82879.	1.1	50
327	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
328	Rare sequence variants in ANO3 and GNAL in a primary torsion dystonia series and controls. Movement Disorders, 2014, 29, 143-147.	2.2	49
329	Metformin Effect on Nontargeted Metabolite Profiles in Patients With Type 2 Diabetes and in Multiple Murine Tissues. Diabetes, 2016, 65, 3776-3785.	0.3	49
330	A genome-wide cross-phenotype meta-analysis of the association of blood pressure with migraine. Nature Communications, 2020, 11, 3368.	5.8	49
331	Genome-wide association analysis in dilated cardiomyopathy reveals two new players in systolic heart failure on chromosomes 3p25.1 and 22q11.23. European Heart Journal, 2021, 42, 2000-2011.	1.0	49
332	Determinants of penetrance and variable expressivity in monogenic metabolic conditions across 77,184 exomes. Nature Communications, 2021, 12, 3505.	5.8	49
333	A functional polymorphism in the SCN1A gene is not associated with carbamazepine dosages in Austrian patients with epilepsy. Epilepsia, 2008, 49, 1108-1109.	2.6	48
334	A meta-analysis of genome-wide data from five European isolates reveals an association of COL22A1, SYT1, and GABRR2with serum creatinine level. BMC Medical Genetics, 2010, 11, 41.	2.1	48
335	Sudden Cardiac Death Due to Deficiency of the Mitochondrial Inorganic Pyrophosphatase PPA2. American Journal of Human Genetics, 2016, 99, 674-682.	2.6	48
336	Gene-based pleiotropy across migraine with aura and migraine without aura patient groups. Cephalalgia, 2016, 36, 648-657.	1.8	47
337	A Low-Frequency Inactivating AKT2 Variant Enriched in the Finnish Population Is Associated With Fasting Insulin Levels and Type 2 Diabetes Risk. Diabetes, 2017, 66, 2019-2032.	0.3	47
338	Congenital heart disease risk loci identified by genome-wide association study in European patients. Journal of Clinical Investigation, 2021, 131, .	3.9	47
339	A genome-wide association meta-analysis on apolipoprotein A-IV concentrations. Human Molecular Genetics, 2016, 25, 3635-3646.	1.4	46
340	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
341	Gene Mapping and Marker Clustering Using Shannon's Mutual Information. IEEE/ACM Transactions on Computational Biology and Bioinformatics, 2006, 3, 47-56.	1.9	44
342	Lack of Association Between the MEF2A Gene and Myocardial Infarction. Circulation, 2008, 117, 185-191.	1.6	44

#	Article	IF	Citations
343	MRPL44 mutations cause a slowly progressive multisystem disease with childhood-onset hypertrophic cardiomyopathy. Neurogenetics, 2015, 16, 319-323.	0.7	44
344	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
345	Expression and mutation analysis of BRUNOL3, a candidate gene for heart and thymus developmental defects associated with partial monosomy 10p. Journal of Molecular Medicine, 2002, 80, 431-442.	1.7	42
346	No evidence for an involvement of variants in the cannabinoid receptor gene (CNR1) in obesity in German children and adolescents. Molecular Genetics and Metabolism, 2007, 90, 429-434.	0.5	42
347	Frequency and Clinical Correlates of Somatic Ying Yang 1 Mutations in Sporadic Insulinomas. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E776-E782.	1.8	42
348	Mutations in PPCS, Encoding Phosphopantothenoylcysteine Synthetase, Cause Autosomal-Recessive Dilated Cardiomyopathy. American Journal of Human Genetics, 2018, 102, 1018-1030.	2.6	42
349	Meta-analysis uncovers genome-wide significant variants for rapid kidney function decline. Kidney International, 2021, 99, 926-939.	2.6	42
350	Mechanisms for multiple intracellular localization of human mitochondrial proteins. Mitochondrion, 2004, 3, 315-325.	1.6	41
351	Immuneâ€Array Analysis in Sporadic Inclusion Body Myositis Reveals HLAâ€“DRB1 Amino Acid Heterogeneity Across the Myositis Spectrum. Arthritis and Rheumatology, 2017, 69, 1090-1099.	2.9	41
352	SCN5A Mutation Type and a Genetic Risk Score Associate Variably With Brugada Syndrome Phenotype in SCN5A Families. Circulation Genomic and Precision Medicine, 2020, 13, e002911.	1.6	41
353	Lack of association of genetic variants in genes of the endocannabinoid system with anorexia nervosa. Child and Adolescent Psychiatry and Mental Health, 2008, 2, 33.	1.2	40
354	Genome-Wide Meta-Analysis of Myopia and Hyperopia Provides Evidence for Replication of 11 Loci. PLoS ONE, 2014, 9, e107110.	1.1	40
355	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
356	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
357	Natriuretic Peptide System Gene Variants Are Associated with Ventricular Dysfunction after Coronary Artery Bypass Grafting. Anesthesiology, 2009, 110, 738-747.	1.3	40
358	Leber’s Hereditary Optic Neuroretinopathy and the X-Chromosomal Susceptibility Factor: No Linkage to DXS7. Human Heredity, 1992, 42, 316-320.	0.4	39
359	Mitochondrial Membrane Protein-Associated Neurodegeneration (MPAN). International Review of Neurobiology, 2013, 110, 73-84.	0.9	39
360	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

#	Article	IF	Citations
361	Restless legs syndrome: Epidemiological and clinicogenetic study in a South Tyrolean population isolate. Movement Disorders, 2006, 21, 1189-1195.	2.2	38
362	Single-Nucleotide Polymorphisms of MMP-2 Gene in Stroke Subtypes. Cerebrovascular Diseases, 2008, 26, 113-119.	0.8	38
363	DYT16 revisited: Exome sequencing identifiesPRKRAmutations in a European dystonia family. Movement Disorders, 2014, 29, 1504-1510.	2.2	38
364	Fifteen Genetic Loci Associated With the Electrocardiographic P Wave. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	38
365	Disentangling the genetics of lean mass. American Journal of Clinical Nutrition, 2019, 109, 276-287.	2.2	38
366	A Population-Based Epidemiological and Genetic Study of X-Linked Retinitis Pigmentosa. , 2007, 48, 4012.		36
367	Preferential reciprocal transfer of paternal/maternal DLK1 alleles to obese children: first evidence of polar overdominance in humans. European Journal of Human Genetics, 2008, 16, 1126-1134.	1.4	36
368	Age-related macular degeneration and functional promoter and coding variants of the apolipoprotein E gene. Human Mutation, 2009, 30, 1048-1053.	1.1	36
369	Infantile Leigh-like syndrome caused by SLC19A3 mutations is a treatable disease. Brain, 2014, 137, e295-e295.	3.7	36
370	DNA methylation and lipid metabolism: an EWAS of 226 metabolic measures. Clinical Epigenetics, 2021, 13, 7.	1.8	36
371	Investigating the causal effect of smoking on hay fever and asthma: a Mendelian randomization meta-analysis in the CARTA consortium. Scientific Reports, 2017, 7, 2224.	1.6	35
372	Lifetime risk of autosomal recessive mitochondrial disorders calculated from genetic databases. EBioMedicine, 2020, 54, 102730.	2.7	35
373	Case-control genetic association study of fibulin-6 (FBLN6orHMCN1) variants in age-related macular degeneration (AMD). Human Mutation, 2007, 28, 406-413.	1.1	34
374	Mitochondrial Genetic Variants Identified to Be Associated with BMI in Adults. PLoS ONE, 2014, 9, e105116.	1.1	34
375	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
376	MRPS22 mutation causes fatal neonatal lactic acidosis with brain and heart abnormalities. Neurogenetics, 2015, 16, 237-240.	0.7	34
377	Genome-wide association study in takotsubo syndrome â€" Preliminary results and future directions. International Journal of Cardiology, 2017, 236, 335-339.	0.8	34
378	Transcriptome-Wide Analysis Identifies Novel Associations With Blood Pressure. Hypertension, 2017, 70, 743-750.	1.3	34

#	Article	IF	Citations
379	Exome sequencing reveals a novel homozygous splice site variant in the WNT1 gene underlying osteogenesis imperfecta type 3. Pediatric Research, 2017, 82, 753-758.	1.1	34
380	Cross-trait analyses with migraine reveal widespread pleiotropy and suggest a vascular component to migraine headache. International Journal of Epidemiology, 2020, 49, 1022-1031.	0.9	34
381	Rare variants in LRRK1 and Parkinson's disease. Neurogenetics, 2014, 15, 49-57.	0.7	33
382	Genome-Wide Genotyping Demonstrates a Polygenic Risk Score Associated With White Matter Hyperintensity Volume in CADASIL. Stroke, 2014, 45, 968-972.	1.0	33
383	Mitochondrial genetic variants identified to be associated with posttraumatic stress disorder. Translational Psychiatry, 2015, 5, e524-e524.	2.4	33
384	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
385	Determinants of occurrence and survival after sudden cardiac arrestâ€“A European perspective: The ESCAPE-NET project. Resuscitation, 2018, 124, 7-13.	1.3	33
386	Genetic Factors Explain a Major Fraction of the 50% Lower Lipoprotein(a) Concentrations in Finns. Arteriosclerosis, Thrombosis, and Vascular Biology, 2018, 38, 1230-1241.	1.1	33
387	Microdeletion syndrome 16p11.2â€p12.2: Clinical and molecular characterization. American Journal of Medical Genetics, Part A, 2009, 149A, 2106-2112.	0.7	32
388	Dilution of candidates: the case of iron-related genes in restless legs syndrome. European Journal of Human Genetics, 2013, 21, 410-414.	1.4	32
389	Concordance of genetic risk across migraine subgroups: Impact on current and future genetic association studies. Cephalalgia, 2015, 35, 489-499.	1.8	32
390	Diverse prevalence of large deletions within the OA1 gene in ocular albinism type 1 patients from Europe and North America. Human Genetics, 2001, 108, 51-54.	1.8	31
391	Family-based association study of the restless legs syndrome loci 2 and 3 in a European population. Movement Disorders, 2007, 22, 207-212.	2.2	31
392	Mapping the Genetic Architecture of Gene Regulation in Whole Blood. PLoS ONE, 2014, 9, e93844.	1.1	31
393	Sequence data and association statistics from 12,940 type 2 diabetes cases and controls. Scientific Data, 2017, 4, 170179.	2.4	31
394	A multi-ancestry genome-wide study incorporating geneâ€“smoking interactions identifies multiple new loci for pulse pressure and mean arterial pressure. Human Molecular Genetics, 2019, 28, 2615-2633.	1.4	31
395	Neurological phenotype and reduced lifespan in heterozygous Tim23 knockout mice, the first mouse model of defective mitochondrial import. Biochimica Et Biophysica Acta - Bioenergetics, 2009, 1787, 371-376.	0.5	30
396	A Candidate Gene Association Study Identifies DAPL1 as a Female-Specific Susceptibility Locus for Age-Related Macular Degeneration (AMD). NeuroMolecular Medicine, 2015, 17, 111-120.	1.8	30

#	Article	IF	Citations
397	Bi-allelic HPDL Variants Cause a Neurodegenerative Disease Ranging from Neonatal Encephalopathy to Adolescent-Onset Spastic Paraplegia. American Journal of Human Genetics, 2020, 107, 364-373.	2.6	30
398	Cohen syndrome diagnosis using whole genome arrays. Journal of Medical Genetics, 2011, 48, 136-140.	1.5	29
399	Compound heterozygous SPATA5 variants in four families and functional studies of SPATA5 deficiency. European Journal of Human Genetics, 2018, 26, 407-419.	1.4	29
400	Genome-wide association meta-analysis of 30,000 samples identifies seven novel loci for quantitative ECG traits. European Journal of Human Genetics, 2019, 27, 952-962.	1.4	29
401	Sequential Defects in Cardiac Lineage Commitment and Maturation Cause Hypoplastic Left Heart Syndrome. Circulation, 2021, 144, 1409-1428.	1.6	29
402	SERAC1 deficiency causes complicated HSP: evidence from a novel splice mutation in a large family. Journal of Medical Genetics, 2018, 55, 39-47.	1.5	28
403	Impairment of Drosophila Orthologs of the Human Orphan Protein C19orf12 Induces Bang Sensitivity and Neurodegeneration. PLoS ONE, 2014, 9, e89439.	1.1	28
404	The Mouse Pink-Eyed Dilution Gene: Association with Hypopigmentation in Prader-Willi and Angelman Syndromes and with Human OCA2. Pigment Cell & Melanoma Research, 1994, 7, 398-402.	4.0	27
405	MITOP: database for mitochondria-related proteins, genes and diseases. Nucleic Acids Research, 1999, 27, 153-155.	6.5	27
406	The common non-synonymous variant G38S of the KCNE1-(minK)-gene is not associated to QT interval in Central European Caucasians: results from the KORA study. European Heart Journal, 2007, 28, 305-309.	1.0	27
407	Genetic variants in RBFOX3 are associated with sleep latency. European Journal of Human Genetics, 2016, 24, 1488-1495.	1.4	27
408	ExomeChip-Wide Analysis of 95 626 Individuals Identifies 10 Novel Loci Associated With QT and JT Intervals. Circulation Genomic and Precision Medicine, 2018, 11, e001758.	1.6	27
409	Effects of Calcium, Magnesium, and Potassium Concentrations on Ventricular Repolarization in Unselected Individuals. Journal of the American College of Cardiology, 2019, 73, 3118-3131.	1.2	27
410	Lowâ€risk variants FGFR2, TNRC9 and LSP1 in German familial breast cancer patients. International Journal of Cancer, 2010, 126, 2858-2862.	2.3	26
411	CRIM1 haploinsufficiency causes defects in eye development in human and mouse. Human Molecular Genetics, 2015, 24, 2267-2273.	1.4	26
412	Neonatal encephalocardiomyopathy caused by mutations in VARS2. Metabolic Brain Disease, 2017, 32, 267-270.	1.4	26
413	Lymphotoxin-Î± and galectin-2 SNPs are not associated with myocardial infarction in two different German populations. Journal of Molecular Medicine, 2007, 85, 997-1004.	1.7	25
414	Severe respiratory complex III defect prevents liver adaptation to prolonged fasting. Journal of Hepatology, 2016, 65, 377-385.	1.8	25

#	Article	IF	Citations
415	Exome Sequencing in Children. Deutsches Ärzteblatt International, 2019, 116, 197-204.	0.6	25
416	Nonviral Glial Cell-Derived Neurotrophic Factor Gene Transfer Enhances Survival of Cultured Dopaminergic Neurons and Improves Their Function after Transplantation in a Rat Model of Parkinsonâ€™s Disease. Human Gene Therapy, 2000, 11, 1529-1541.	1.4	24
417	Mutation screen and association studies in the Diacylglycerol O-acyltransferase homolog 2 gene (DGAT2), a positional candidate gene for early onset obesity on chromosome 11q13. BMC Genetics, 2007, 8, 17.	2.7	24
418	The Role of Biobanking in Rare Diseases: European Consensus Expert Group Report. Biopreservation and Biobanking, 2009, 7, 155-156.	0.5	24
419	<scp>PSEA</scp>: Phenotype Set Enrichment Analysisâ€”A New Method for Analysis of Multiple Phenotypes. Genetic Epidemiology, 2012, 36, 244-252.	0.6	24
420	Replication study of multiple sclerosis (MS) susceptibility alleles and correlation of DNA-variants with disease features in a cohort of Austrian MS patients. Neurogenetics, 2012, 13, 181-187.	0.7	24
421	Simulation of Finnish Population History, Guided by Empirical Genetic Data, to Assess Power of Rare-Variant Tests in Finland. American Journal of Human Genetics, 2014, 94, 710-720.	2.6	24
422	Genome-wide association study for refractive astigmatism reveals genetic co-determination with spherical equivalent refractive error: the CREAM consortium. Human Genetics, 2015, 134, 131-146.	1.8	24
423	NFAT5 and SLC4A10 Loci Associate with Plasma Osmolality. Journal of the American Society of Nephrology: JASN, 2017, 28, 2311-2321.	3.0	24
424	Rare coding variants in 35 genes associate with circulating lipid levelsâ€”A multi-ancestry analysis of 170,000 exomes. American Journal of Human Genetics, 2022, 109, 81-96.	2.6	24
425	Mapping of Xp21 translocation breakpoints in and around the DMD gene by pulsed field gel electrophoresis. Genomics, 1988, 3, 315-322.	1.3	23
426	Association of variants in the BAT1-NFKBIL1-LTA genomic region with protection against myocardial infarction in Europeans. Human Molecular Genetics, 2007, 16, 1821-1827.	1.4	23
427	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
428	Lessons from exome sequencing in prenatally diagnosed heart defects: A basis for prenatal testing. Clinical Genetics, 2019, 95, 582-589.	1.0	23
429	SQSTM1/p62 variants in 486 patients with familial ALS from Germany and Sweden. Neurobiology of Aging, 2020, 87, 139.e9-139.e15.	1.5	23
430	Common and rare susceptibility genetic variants predisposing to Brugada syndrome in Thailand. Heart Rhythm, 2020, 17, 2145-2153.	0.3	23
431	Bayesian and frequentist analysis of an Austrian genome-wide association study of colorectal cancer and advanced adenomas. Oncotarget, 2017, 8, 98623-98634.	0.8	23
432	Sequence diversity ofKIAA0027/MLC1:are megalencephalic leukoencephalopathy and schizophrenia allelic disorders?. Human Mutation, 2003, 21, 45-52.	1.1	22

#	Article	IF	Citations
433	Mutations in the CLCN2 gene are a rare cause of idiopathic generalized epilepsy syndromes. Neurogenetics, 2006, 7, 265-268.	0.7	22
434	Genes predict village of origin in rural Europe. European Journal of Human Genetics, 2010, 18, 1269-1270.	1.4	22
435	Lack of replication in polymorphisms reported to be associated with atrial fibrillation. Heart Rhythm, 2011, 8, 403-409.	0.3	22
436	Growth hormone (GH), insulin-like growth factors (IGFs), and IGF-binding protein-3 (IGFBP-3) in a child with Proteus syndrome. American Journal of Medical Genetics Part A, 1994, 50, 204-210.	2.4	21
437	The Gene Coding for Glial Cell Line Derived Neurotrophic Factor (GDNF) Maps to Chromosome 5p12-p13.1. Genomics, 1995, 28, 605-607.	1.3	21
438	Retinal detachment and cataract, facial dysmorphism, generalized osteoporosis, immobile spine and platyspondyly in a consanguinous kindred - a possible new syndrome. Clinical Genetics, 2001, 59, 99-105.	1.0	21
439	Genetic Modifiers for the Long-QT Syndrome. Circulation: Cardiovascular Genetics, 2016, 9, 330-339.	5.1	21
440	A novelLRRK2 mutation in an Austrian cohort of patients with Parkinson's disease. Movement Disorders, 2007, 22, 1640-1643.	2.2	20
441	Genetic Variation in the Lymphotoxin-Alpha Pathway and the Risk of Ischemic Stroke in European Populations. Stroke, 2009, 40, 970-972.	1.0	20
442	The many faces of paediatric mitochondrial disease on neuroimaging. Child's Nervous System, 2016, 32, 2077-2083.	0.6	20
443	Screening forCHCHD10mutations in a large cohort of sporadic ALS patients: no evidence for pathogenicity of the p.P34S variant: Table 1. Brain, 2016, 139, e8-e8.	3.7	20
444	Effect of Insulin Resistance on Monounsaturated Fatty Acid Levels: A Multi-cohort Non-targeted Metabolomics and Mendelian Randomization Study. PLoS Genetics, 2016, 12, e1006379.	1.5	20
445	3.7ÂMb tandem microduplication in chromosome 5p13.1-p13.2 associated withÂdevelopmental delay, macrocephaly, obesity, and lymphedema. Further characterization of the dup(5p13) syndrome. European Journal of Medical Genetics, 2011, 54, 225-230.	0.7	19
446	Common and Rare Coding Genetic Variation Underlying the Electrocardiographic PR Interval. Circulation Genomic and Precision Medicine, 2018, 11, e002037.	1.6	19
447	Sequence variants in four genes underlying Bardet-Biedl syndrome in consanguineous families. Molecular Vision, 2017, 23, 482-494.	1.1	19
448	A Gene Locus Responsible for the Familial Hair Shaft Abnormality Pili Annulati Maps to Chromosome 12q24.32â€“24.33. Journal of Investigative Dermatology, 2004, 123, 1073-1077.	0.3	18
449	Habitual sleep disturbances and migraine: a Mendelian randomization study. Annals of Clinical and Translational Neurology, 2020, 7, 2370-2380.	1.7	18
450	A multi-ethnic epigenome-wide association study of leukocyte DNA methylation and blood lipids. Nature Communications, 2021, 12, 3987.	5.8	18

#	Article	IF	Citations
451	Blood DNA methylation provides an accurate biomarker of KMT2B-related dystonia and predicts onset. Brain, 2022, 145, 644-654.	3.7	18
452	Genetic loci and prioritization of genes for kidney function decline derived from a meta-analysis of 62 longitudinal genome-wide association studies. Kidney International, 2022, 102, 624-639.	2.6	18
453	Linkage of X-linked retinitis pigmentosa to the hypervariable DNA marker M27? (DXS255). Human Genetics, 1989, 81, 283-6.	1.8	17
454	Deletion analysis maps ocular albinism proximal to the steroid sulphatase locus. Clinical Genetics, 1993, 43, 169-173.	1.0	17
455	Genetic Structure in Contemporary South Tyrolean Isolated Populations Revealed by Analysis of Y-Chromosome, mtDNA, and Alu Polymorphisms. Human Biology, 2006, 78, 441-464.	0.4	17
456	MTO1-Deficient Mouse Model Mirrors the Human Phenotype Showing Complex I Defect and Cardiomyopathy. PLoS ONE, 2014, 9, e114918.	1.1	17
457	Associations between calcium and vitamin D supplement use as well as their serum concentrations and subclinical cardiovascular disease phenotypes. Atherosclerosis, 2015, 241, 743-751.	0.4	17
458	A biallelic mutation linksMYORGto autosomal-recessive primary familial brain calcification. Brain, 2019, 142, e4-e4.	3.7	17
459	Mendelian randomization evaluation of causal effects of fibrinogen on incident coronary heart disease. PLoS ONE, 2019, 14, e0216222.	1.1	17
460	Gene-educational attainment interactions in a multi-ancestry genome-wide meta-analysis identify novel blood pressure loci. Molecular Psychiatry, 2020, 26, 2111-2125.	4.1	17
461	Bi-allelic Variants in RALGAPA1 Cause Profound Neurodevelopmental Disability, Muscular Hypotonia, Infantile Spasms, and Feeding Abnormalities. American Journal of Human Genetics, 2020, 106, 246-255.	2.6	17
462	Genome-wide meta-analysis of phytosterols reveals five novel loci and a detrimental effect on coronary atherosclerosis. Nature Communications, 2022, 13, 143.	5.8	17
463	Spondylo-ocular syndrome: a new entity with crystalline lens malformation, cataract, retinal detachment, osteoporosis, and platyspondyly. American Journal of Ophthalmology, 2003, 135, 681-687.	1.7	16
464	A genomic exploration identifies mechanisms that may explain adverse cardiovascular effects of COX-2 inhibitors. Scientific Reports, 2017, 7, 10252.	1.6	16
465	Diagnostic exome sequencing in non-acquired focal epilepsies highlights a major role of GATOR1 complex genes. Journal of Medical Genetics, 2020, 57, 624-633.	1.5	16
466	Bi-allelic truncating mutations in VWA1 cause neuromyopathy. Brain, 2021, 144, 574-583.	3.7	16
467	STAT3 single-nucleotide polymorphisms and STAT3 mutations associated with hyper-IgE syndrome are not responsible for increased serum IgE serum levels in asthma families. European Journal of Human Genetics, 2009, 17, 352-356.	1.4	15
468	Genome wide association study identifies variants in NBEA associated with migraine in bipolar disorder. Journal of Affective Disorders, 2015, 172, 453-461.	2.0	15

#	Article	IF	Citations
469	Gene-gene Interaction Analyses for Atrial Fibrillation. Scientific Reports, 2016, 6, 35371.	1.6	15
470	EARS2 mutations cause fatal neonatal lactic acidosis, recurrent hypoglycemia and agenesis of corpus callosum. Metabolic Brain Disease, 2016, 31, 717-721.	1.4	15
471	Stimulation of soluble guanylyl cyclase (sGC) by riociguat attenuates heart failure and pathological cardiac remodelling. British Journal of Pharmacology, 2022, 179, 2430-2442.	2.7	15
472	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
473	MicroRNA-365 regulates human cardiac action potential duration. Nature Communications, 2022, 13, 220.	5.8	15
474	Common Genetic Variants Contribute to Risk of Transposition of the Great Arteries. Circulation Research, 2022, 130, 166-180.	2.0	15
475	Cloning and gene structure of the rod cGMP phosphodiesterase delta subunit gene (PDED) in man and mouse. European Journal of Human Genetics, 1998, 6, 283-290.	1.4	14
476	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
477	A homozygous splice variant in AP4S1 mimicking neurodegeneration with brain iron accumulation. Movement Disorders, 2017, 32, 797-799.	2.2	14
478	Identification of a Novel Heterozygous De Novo 7-bp Frameshift Deletion in PBX1 by Whole-Exome Sequencing Causing a Multi-Organ Syndrome Including Bilateral Dysplastic Kidneys and Hypoplastic Clavicles. Frontiers in Pediatrics, 2017, 5, 251.	0.9	14
479	Biallelic Mutations in SLC1A2; an Additional Mode of Inheritance for SLC1A2-Related Epilepsy. Neuropediatrics, 2018, 49, 059-062.	0.3	14
480	Association of alcohol consumption with allergic disease and asthma: a multiâ€centre Mendelian randomization analysis. Addiction, 2019, 114, 216-225.	1.7	14
481	Cis-epistasis at the LPA locus and risk of cardiovascular diseases. Cardiovascular Research, 2022, 118, 1088-1102.	1.8	14
482	Evidence for genetic heterogeneity in Best's vitelliform macular dystrophy.. Journal of Medical Genetics, 1995, 32, 855-858.	1.5	13
483	Pantothenate Kinase-Associated Neurodegeneration. Current Drug Targets, 2012, 13, 1182-1189.	1.0	13
484	Coexisting variants in OSTM1 and MANEAL cause a complex neurodegenerative disorder with NBIA-like brain abnormalities. European Journal of Human Genetics, 2017, 25, 1092-1095.	1.4	13
485	Ellisâ€“van Creveld syndrome and profound deafness resulted by sequence variants in the EVCÂ/ÂEVC2Âand TMC1Âgenes. Journal of Genetics, 2017, 96, 1005-1014.	0.4	13
486	Multi-ancestry genome-wide geneâ€“sleep interactions identify novel loci for blood pressure. Molecular Psychiatry, 2021, 26, 6293-6304.	4.1	13

#	Article	IF	Citations
487	X-linked ocular albinism (Nettleship-Falls): a novel 29-bp deletion in exon 1.. , 2001, 239, 167-172.		12
488	Clinical Picture, Evolution and Peculiar Molecular Findings in a Very Large Pedigree with Wolfram Syndrome. Journal of Pediatric Endocrinology and Metabolism, 2005, 18, 1391-7.	0.4	12
489	Generation of a human induced pluripotent stem cell (iPSC) line from a patient carrying a P33T mutation in the PDX1 gene. Stem Cell Research, 2016, 17, 273-276.	0.3	12
490	Generation of a human induced pluripotent stem cell (iPSC) line from a patient with family history of diabetes carrying a C18R mutation in the PDX1 gene. Stem Cell Research, 2016, 17, 292-295.	0.3	12
491	A new missense mutation in UMOD gene leads to severely reduced serum uromodulin concentrations â€" A tool for the diagnosis of uromodulin-associated kidney disease. Clinical Biochemistry, 2017, 50, 155-158.	0.8	12
492	De novo stop-loss variants in CLDN11 cause hypomyelinating leukodystrophy. Brain, 2021, 144, 411-419.	3.7	12
493	Defective phosphatidylethanolamine biosynthesis leads to a broad ataxia-spasticity spectrum. Brain, 2021, 144, e30-e30.	3.7	12
494	Familial carpal tunnel syndrome: further evidence for a genetic contribution. Clinical Genetics, 2005, 69, 179-182.	1.0	11
495	Systematic mutation analysis of KIAA0767 and KIAA1646 in chromosome 22q-linked periodic catatonia. BMC Psychiatry, 2005, 5, 36.	1.1	11
496	Anticholinergicâ€responsive gait freezing in a patient with pantothenate kinaseâ€associated neurodegeneration. Movement Disorders, 2008, 23, 283-284.	2.2	11
497	How to link call rate and pâ€values for Hardyâ€"Weinberg equilibrium as measures of genomeâ€wide SNP data quality. Statistics in Medicine, 2010, 29, 2347-2358.	0.8	11
498	A De Novo Missense Variant in POU3F2 Identified in a Child with Global Developmental Delay. Neuropediatrics, 2018, 49, 401-404.	0.3	11
499	PRUNE1 Deficiency: Expanding the Clinical and Genetic Spectrum. Neuropediatrics, 2018, 49, 330-338.	0.3	11
500	Identification of disease-causing variants by comprehensive genetic testing with exome sequencing in adults with suspicion of hereditary FSGS. European Journal of Human Genetics, 2021, 29, 262-270.	1.4	11
501	Bi-allelic variants in SPATA5L1 lead to intellectual disability, spastic-dystonic cerebral palsy, epilepsy, and hearing loss. American Journal of Human Genetics, 2021, 108, 2006-2016.	2.6	11
502	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
503	Lifetime risk of autosomal recessive neurodegeneration with brain iron accumulation (NBIA) disorders calculated from genetic databases. EBioMedicine, 2022, 77, 103869.	2.7	11
504	Generation and characterization of radiation reduced cell hybrids and isolation of probes from the proximal short arm of the human X chromosome. Human Genetics, 1992, 90, 243-246.	1.8	10

#	Article	IF	Citations
505	Do common genetic variants in endotoxin signaling pathway contribute to predisposition to alcoholic liver cirrhosis?. Clinical Chemistry and Laboratory Medicine, 2009, 47, 398-404.	1.4	10
506	Genetic Variants Associated With Atrial Fibrillation and PR Interval Following Cardiac Surgery. Journal of Cardiothoracic and Vascular Anesthesia, 2015, 29, 605-610.	0.6	10
507	Response by Crotti et al to Letter Regarding Article, â€œGenetic Modifiers for the Long-QT Syndrome: How Important Is the Role of Variants in the 3â€² Untranslated Region of KCNQ1?â€• Circulation: Cardiovascular Genetics, 2016, 9, 581-582.	5.1	10
508	Biallelic lossâ€ofâ€function variants inRBL2in siblings with a neurodevelopmental disorder. Annals of Clinical and Translational Neurology, 2020, 7, 390-396.	1.7	10
509	Rescue of STAT3 Function in Hyper-IgE Syndrome Using Adenine Base Editing. CRISPR Journal, 2021, 4, 178-190.	1.4	10
510	Identification of a de novo microdeletion 1q44 in a patient with hypogenesis of the corpus callosum, seizures and microcephaly â€“ A case report. Gene, 2017, 616, 41-44.	1.0	9
511	MAP2 â€“ A Candidate Gene for Epilepsy, Developmental Delay and Behavioral Abnormalities in a Patient With Microdeletion 2q34. Frontiers in Genetics, 2018, 9, 99.	1.1	9
512	Fatal metabolic decompensation in carbonic anhydrase VA deficiency despite early treatment and control of hyperammonemia. Genetics in Medicine, 2020, 22, 654-655.	1.1	9
513	Serum uromodulin is inversely associated with biomarkers of subclinical inflammation in the population-based KORA F4 study. CKJ: Clinical Kidney Journal, 2021, 14, 1618-1625.	1.4	9
514	The breakpoint identified in a balanced de novo translocation t(7;9)(p14.1;q31.3) disrupts the A-kinase (PRKA) anchor protein 2 gene (AKAP2) on chromosome 9 in a patient with Kallmann syndrome and bone anomalies. International Journal of Molecular Medicine, 2007, 19, 429-35.	1.8	9
515	Three novel mutations (I506S, S466X, 1651Aâ†'T) in exon 10 of the cystic fibrosis transmembrane conductance regulator (CFTR) detected in patients of Southern German Descent. Human Mutation, 1994, 3, 64-66.	1.1	8
516	A familial deletion in the Prader-Willi syndrome region including the imprinting control region. , 1996, 8, 288-292.		8
517	A genome-wide analysis of DNA methylation identifies a novel association signal for Lp(a) concentrations in the LPA promoter. PLoS ONE, 2020, 15, e0232073.	1.1	8
518	Mutation analysis in the diagnosis of cystic fibrosis. European Journal of Pediatrics, 1993, 152, 909-911.	1.3	7
519	In frame deletion (Î”F311) within a short trinucleotide repeat of the first transmembrane region of the cystic fibrosis gene. Human Molecular Genetics, 1993, 2, 2173-2174.	1.4	7
520	A Systematic Evaluation of Short Tandem Repeats in Lipid Candidate Genes: Riding on the SNP-Wave. PLoS ONE, 2014, 9, e102113.	1.1	7
521	Biallelic mutations in PIGP cause developmental and epileptic encephalopathy. Annals of Clinical and Translational Neurology, 2019, 6, 968-973.	1.7	7
522	Wholeâ€exome sequencing revealed a nonsense mutation in STKLD1 causing nonâ€syndromic preâ€axial polydactyly type A affecting only upper limb. Clinical Genetics, 2019, 96, 134-139.	1.0	7

#	Article	IF	Citations
523	Elucidating the relationship between migraine risk and brain structure using genetic data. Brain, 2022, 145, 3214-3224.	3.7	7
524	Clonal hematopoiesis as a pitfall in germline variant interpretation in the context of Mendelian disorders. Human Molecular Genetics, 2022, 31, 2386-2395.	1.4	7
525	A multimetric approach to analysis of genome-wide association by single markers and composite likelihood. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 2592-2597.	3.3	6
526	Anal atresia, coloboma, microphthalmia, and nasal skin tag in a female patient with 3.5â€‰Mb deletion of 3q26 encompassing SOX2. American Journal of Medical Genetics, Part A, 2013, 161, 1421-1424.	0.7	6
527	Low-frequency variation near common germline susceptibility loci are associated with risk of Ewing sarcoma. PLoS ONE, 2020, 15, e0237792.	1.1	6
528	A novel pathogenic variant in MYO18B associating early-onset muscular hypotonia, and characteristic dysmorphic features, delineation of the phenotypic spectrum of MYO18B-related conditions. Gene, 2020, 742, 144542.	1.0	6
529	Delineation of epileptic and neurodevelopmental phenotypes associated with variants in STX1B. Seizure: the Journal of the British Epilepsy Association, 2021, 87, 25-29.	0.9	6
530	Identification of a Functional PDE5A Variant at the Chromosome 4q27 Coronary Artery Disease Locus in an Extended Myocardial Infarction Family. Circulation, 2021, 144, 662-665.	1.6	6
531	TaqMan assays for genotyping of single nucleotide polymorphisms present at a disease susceptibility locus on chromosome 6. Clinical Chemistry and Laboratory Medicine, 2005, 43, 167-72.	1.4	5
532	Genetic Structure in Contemporary South Tyrolean Isolated Populations Revealed by Analysis of Y-Chromosome, mtDNA, and Alu Polymorphisms. Human Biology, 2009, 81, 875-898.	0.4	5
533	Variants in STAT5B Associate with Serum TC and LDL-C Levels. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E1496-E1501.	1.8	5
534	Somatic alterations compromised molecular diagnosis of DOCK8 hyper-IgE syndrome caused by a novel intronic splice site mutation. Scientific Reports, 2018, 8, 16719.	1.6	5
535	Extensive Natural Variation for Cellular Hydrogen Peroxide Release Is Genetically Controlled. PLoS ONE, 2012, 7, e43566.	1.1	5
536	Genomic correlates of glatiramer acetate adverse cardiovascular effects lead to a novel locus mediating coronary risk. PLoS ONE, 2017, 12, e0182999.	1.1	5
537	Association of circulating MR-proADM with all-cause and cardiovascular mortality in the general population: Results from the KORA F4 cohort study. PLoS ONE, 2022, 17, e0262330.	1.1	5
538	Genetic and clinical determinants of abdominal aortic diameter: genome-wide association studies, exome array data and Mendelian randomization study. Human Molecular Genetics, 2022, 31, 3566-3579.	1.4	5
539	Recent advances in the genetics of atrial fibrillation: from rare and common genetic variants to microRNA signaling. Neurology International, 2011, 1, .	0.2	4
540	Adult-onset variant ataxia-telangiectasia diagnosed by exome and cDNA sequencing. Neurology: Genetics, 2019, 5, e346.	0.9	4

#	Article	IF	Citations
541	Phenotypic variability of GABRA1â€related epilepsy in monozygotic twins. Annals of Clinical and Translational Neurology, 2019, 6, 2317-2322.	1.7	4
542	A homozygous truncating variant in CCDC186 in an individual with epileptic encephalopathy. Annals of Clinical and Translational Neurology, 2021, 8, 278-283.	1.7	4
543	TP53 germline mutations in the context of families with hereditary breast and ovarian cancer: a clinical challenge. Archives of Gynecology and Obstetrics, 2021, 303, 1557-1567.	0.8	4
544	Defining the genotypic and phenotypic spectrum of X-linked MSL3-related disorder. Genetics in Medicine, 2021, 23, 384-395.	1.1	4
545	A comprehensive phenotypic characterization of a whole-body Wdr45 knock-out mouse. Mammalian Genome, 2021, 32, 332-349.	1.0	4
546	Genome-Wide Analyses Identify KIF5A as a Novel ALS Gene. SSRN Electronic Journal, 0, , .	0.4	4
547	Identification of a novel missense mutation (G314E) in exon 7 of the cystic fibrosis transmembrane conductance regulator gene identified in a CF patient with pancreatic sufficiency. Human Mutation, 1994, 3, 67-68.	1.1	3
548	Procolipase Gene: No Association with Early-Onset Obesity or Fat Intake. Obesity Facts, 2009, 2, 40-44.	1.6	3
549	Recurrent spontaneous coronary dissections in a patient with a de novo fibrillin-1 mutation without Marfan syndrome. Thrombosis and Haemostasis, 2015, 113, 668-670.	1.8	3
550	Biotinidase deficiency. Neurology: Genetics, 2020, 6, e525.	0.9	3
551	Variants associated with HHIP expression have sex-differential effects on lung function. Wellcome Open Research, 2020, 5, 111.	0.9	3
552	MATR3 haploinsufficiency and early-onset neurodegeneration. Brain, 2021, 144, e72-e72.	3.7	3
553	Chronic recurrent multifocal osteomyelitis (CRMO): evidence for a susceptibility gene located on chromosome 18q21.3-18q22. , 0, .		3
554	Suleiman-El-Hattab syndrome: a histone modification disorder caused by TASP1 deficiency. Human Molecular Genetics, 2022, 31, 3083-3094.	1.4	3
555	Widening the view. Nature Genetics, 1997, 15, 224-225.	9.4	2
556	Periphilin is a novel interactor of synphilin-1, a protein implicated in Parkinson's disease. Neurogenetics, 2010, 11, 203-215.	0.7	2
557	Sampling GWAS subjects from risk populations. Genetic Epidemiology, 2011, 35, 148-153.	0.6	2
558	Multi-ancestry genome-wide association study accounting for gene-psychosocial factor interactions identifies novel loci for blood pressure traits. Human Genetics and Genomics Advances, 2021, 2, 100013.	1.0	2

#	Article	IF	Citations
559	Blood cis-eQTL Analysis Fails to Identify Novel Association Signals among Sub-Threshold Candidates from Genome-Wide Association Studies in Restless Legs Syndrome. PLoS ONE, 2014, 9, e98092.	1.1	2
560	Reduced amplification efficiency of KIAA0027/MLC1 alleles: implications for the molecular diagnosis of megalencephalic leukoencephalopathy with subcortical cysts. Molecular and Cellular Probes, 2002, 16, 379-384.	0.9	1
561	Absence of BiP Co-chaperone DNAJC3 Causes Diabetes Mellitus and Multisystemic Neurodegeneration. American Journal of Human Genetics, 2015, 96, 514.	2.6	1
562	Response to Comment on Adam et al. Metformin Effect on Nontargeted Metabolite Profiles in Patients With Type 2 Diabetes and in Multiple Murine Tissues. Diabetes 2016;65:3776â€“3785. Diabetes, 2017, 66, e3-e4.	0.3	1
563	pulver: an R package for parallel ultra-rapid p-value computation for linear regression interaction terms. BMC Bioinformatics, 2017, 18, 429.	1.2	1
564	Generation of a human induced pluripotent stem cell line (HMGUi002-A) from a healthy male individual. Stem Cell Research, 2019, 39, 101531.	0.3	1
565	Reply: Adult-onset distal spinal muscular atrophy: a new phenotype associated with KIF5A mutations. Brain, 2019, 142, e67-e67.	3.7	1
566	Abstract 12059: Incidental Findings in Cardiomyopathy and Channelopathy Genes Among 5891 Individuals Undergoing Whole-exome Sequencing. What Should be Reported?. Circulation, 2015, 132, .	1.6	1
567	Exome sequencing in individuals with cardiovascular laterality defects identifies potential candidate genes. European Journal of Human Genetics, 2022, , .	1.4	1
568	Report of the Second International Workshop on Human Chromosome 10 Mapping 1997. Cytogenetic and Genome Research, 1997, 78, 183-196.	0.6	0
569	Correction: The MITOP database â€“ a one-stop shop for mitochondrial information. Trends in Genetics, 1999, 15, 82.	2.9	0
570	Cryptic del/dup aberration of 60.6ÂMb at 5q15-5q23.3 predicting adult-onset leukodystrophy. European Journal of Medical Genetics, 2012, 55, 568-572.	0.7	0
571	Exomdiagnostik verÃndert die Sicht auf Mitochondriopathien. Medizinische Genetik, 2012, 24, 183-186.	0.1	0
572	Does B-type Natriuretic Peptide or Its Gene Polymorphism Predict Patient Outcome after Coronary Artery Bypass Graft Surgery?. Anesthesiology, 2009, 111, 1378-1379.	1.3	0
573	Identification of Recurring Tumor-Specific Somatic Mutations In Acute Myeloid Leukemia by Transcriptome Sequencing.. Blood, 2010, 116, 1059-1059.	0.6	0
574	The Norrie Disease Gene: Positional Cloning, Mutation Analysis and Protein Homologies. , 1993, , 135-142.		0
575	LRRK2 (Leucine-Rich Repeat Kinase 2) Gene on PARK8 Locus in Families with Parkinsonism. , 2008, , 75-89.		0
576	Effect of genome-wide simultaneous hypotheses tests on the discovery rate. International Journal of Molecular Epidemiology and Genetics, 2011, 2, 163-77.	0.4	0

#	Article	IF	Citations
577	Title is missing!. , 2020, 15, e0237792.		0
578	Title is missing!. , 2020, 15, e0237792.		0
579	Title is missing!. , 2020, 15, e0237792.		0
580	Title is missing!. , 2020, 15, e0237792.		0
581	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