A general framework for estimating the relative pathog

Nature Genetics

46, 310-315

DOI: 10.1038/ng.2892

Citation Report

#	Article	IF	Citations
1	Harnessing Gene Expression Networks to Prioritize Candidate Epileptic Encephalopathy Genes. PLoS ONE, 2014, 9, e102079.	1.1	25
2	Evidence for Evolutionary and Nonevolutionary Forces Shaping the Distribution of Human Genetic Variants near Transcription Start Sites. PLoS ONE, 2014, 9, e114432.	1.1	4
3	Architecture of Inherited Susceptibility to Colorectal Cancer: A Voyage of Discovery. Genes, 2014, 5, 270-284.	1.0	13
4	Targeted nanotherapeutics in cancer. International Journal of Nanomedicine, 2014, 9, 1627.	3.3	7
5	Adaptors for disorders of the brain? The cancer signaling proteins NEDD9, CASS4, and PTK2B in Alzheimerâ€™s disease. Oncoscience, 2014, 1, 486-503.	0.9	38
6	A Test for Ancient Selective Sweeps and an Application to Candidate Sites in Modern Humans. Molecular Biology and Evolution, 2014, 31, 3344-3358.	3.5	36
7	In silico prediction of splice-altering single nucleotide variants in the human genome. Nucleic Acids Research, 2014, 42, 13534-13544.	6.5	396
8	The Challenge of Next Generation Sequencing in the Context of Neuromuscular Diseases. Journal of Neuromuscular Diseases, 2014, 1, 135-149.	1.1	25
9	Approaches for establishing the function of regulatory genetic variants involved in disease. Genome Medicine, 2014, 6, 92.	3.6	34
10	OncoCis: annotation of cis-regulatory mutations in cancer. Genome Biology, 2014, 15, 485.	3.8	22
11	FunSeq2: a framework for prioritizing noncoding regulatory variants in cancer. Genome Biology, 2014, 15, 480.	3.8	291
12	VariantDB: a flexible annotation and filtering portal for next generation sequencing data. Genome Medicine, 2014, 6, 74.	3.6	60
13	VAS: a convenient web portal for efficient integration of genomic features with millions of genetic variants. BMC Genomics, 2014, 15, 886.	1.2	1
14	FLAGS, frequently mutated genes in public exomes. BMC Medical Genomics, 2014, 7, 64.	0.7	108
15	Walking the interactome for candidate prioritization in exome sequencing studies of Mendelian diseases. Bioinformatics, 2014, 30, 3215-3222.	1.8	91
16	Identification of Rare Causal Variants in Sequence-Based Studies: Methods and Applications to VPS13B, a Gene Involved in Cohen Syndrome and Autism. PLoS Genetics, 2014, 10, e1004729.	1.5	45
17	Analysis of Stop-Gain and Frameshift Variants in Human Innate Immunity Genes. PLoS Computational Biology, 2014, 10, e1003757.	1.5	32
18	Approximation to the Distribution of Fitness Effects across Functional Categories in Human Segregating Polymorphisms. PLoS Genetics, 2014, 10, e1004697.	1.5	59

#	Article	IF	Citations
19	The road from next-generation sequencing to personalized medicine. Personalized Medicine, 2014, 11, 523-544.	0.8	40
20	RNF213 Rare Variants in an Ethnically Diverse Population With Moyamoya Disease. Stroke, 2014, 45, 3200-3207.	1.0	129
21	Analysis of the ABCA4 genomic locus in Stargardt disease. Human Molecular Genetics, 2014, 23, 6797-6806.	1.4	117
22	The transcriptional regulator ADNP links the BAF (SWI/SNF) complexes with autism. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 315-326.	0.7	68
23	Single nucleotide variations: Biological impact and theoretical interpretation. Protein Science, 2014, 23, 1650-1666.	3.1	94
24	Genetic associations of nonsynonymous exonic variants with psychophysiological endophenotypes. Psychophysiology, 2014, 51, 1300-1308.	1.2	21
25	Medical genomics: The intricate path from genetic variant identification to clinical interpretation. Applied & Translational Genomics, 2014, 3, 60-67.	2.1	32
26	Transcriptome Sequencing of a Large Human Family Identifies the Impact of Rare Noncoding Variants. American Journal of Human Genetics, 2014, 95, 245-256.	2.6	63
27	Integrated Sequence Analysis Pipeline Provides One-Stop Solution for Identifying Disease-Causing Mutations. Human Mutation, 2014, 35, 1427-1435.	1.1	31
28	Exome sequencing and genome-wide copy number variant mapping reveal novel associations with sensorineural hereditary hearing loss. BMC Genomics, 2014, 15, 1155.	1.2	27
29	Analysis for co-occurring sequence features identifies link between common synonymous variant and an early-terminated NPC1 isoform. Journal of Clinical Bioinformatics, 2014, 4, 14.	1.2	0
30	SuRFing the genomics wave: an R package for prioritising SNPs by functionality. Genome Medicine, 2014, 6, 79.	3.6	15
31	Computational approaches to interpreting genomic sequence variation. Genome Medicine, 2014, 6, 87.	3.6	33
32	The National Institutes of Health undiagnosed diseases program. Current Opinion in Pediatrics, 2014, 26, 626-633.	1.0	43
33	Hoyeraal-Hreidarsson syndrome caused by a germline mutation in the TEL patch of the telomere protein TPP1. Genes and Development, 2014, 28, 2090-2102.	2.7	106
34	Elucidating the Genotypeâ€“Phenotype Relationships and Network Perturbations of Human Shared and Specific Disease Genes from an Evolutionary Perspective. Genome Biology and Evolution, 2014, 6, 2741-2753.	1.1	6
35	Whole exome or genome sequencing: nurses need to prepare families for the possibilities. Journal of Advanced Nursing, 2014, 70, 2736-2745.	1.5	8
36	A Higher Mutational Burden in Females Supports a â€œFemale Protective Modelâ€•in Neurodevelopmental Disorders. American Journal of Human Genetics, 2014, 94, 415-425.	2.6	457

#	Article	IF	Citations
38	Prioritization of neurodevelopmental disease genes by discovery of new mutations. Nature Neuroscience, 2014, 17, 764-772.	7.1	148
39	Guidelines for genetic studies in single patients: lessons from primary immunodeficiencies. Journal of Experimental Medicine, 2014, 211, 2137-2149.	4.2	218
40	Characteristics of Neutral and Deleterious Protein-Coding Variation among Individuals and Populations. American Journal of Human Genetics, 2014, 95, 421-436.	2.6	89
41	Rare and low-frequency variants in human common diseases and other complex traits. Journal of Medical Genetics, 2014, 51, 705-714.	1.5	29
42	pENCODE: A Plant Encyclopedia of DNA Elements. Annual Review of Genetics, 2014, 48, 49-70.	3.2	38
43	Mutations in SLC13A5 Cause Autosomal-Recessive Epileptic Encephalopathy with Seizure Onset in the First Days of Life. American Journal of Human Genetics, 2014, 95, 113-120.	2.6	112
44	Pathogenic Variants for Mendelian and Complex Traits in Exomes of 6,517 European and African Americans: Implications for the Return of Incidental Results. American Journal of Human Genetics, 2014, 95, 183-193.	2.6	78
45	MutaCYP: Classification of missense mutations in human cytochromes P450. BMC Medical Genomics, 2014, 7, 47.	0.7	8
46	Hotspot activating PRKD1 somatic mutations in polymorphous low-grade adenocarcinomas of the salivary glands. Nature Genetics, 2014, 46, 1166-1169.	9.4	188
47	Saturation editing of genomic regions by multiplex homology-directed repair. Nature, 2014, 513, 120-123.	13.7	301
48	All together now for variant interpretation. Nature Reviews Genetics, 2014, 15, 216-216.	7.7	1
49	Variant Association Tools for Quality Control and Analysis of Large-Scale Sequence and Genotyping Array Data. American Journal of Human Genetics, 2014, 94, 770-783.	2.6	71
50	Review of Current Methods, Applications, and Data Management for the Bioinformatics Analysis of Whole Exome Sequencing. Cancer Informatics, 2014, 13s2, CIN.S13779.	0.9	113
51	Functional Annotation of Putative Regulatory Elements at Cancer Susceptibility Loci. Cancer Informatics, 2014, 13s2, CIN.S13789.	0.9	6
52	Targeted high-throughput sequencing for genetic diagnostics of hemophagocytic lymphohistiocytosis. Genome Medicine, 2015, 7, 130.	3.6	37
53	Two novel mutations in ILDR1 gene cause autosomal recessive nonsyndromic hearing loss in consanguineous Iranian families. Journal of Genetics, 2015, 94, 483-487.	0.4	14
54	Evaluation of CADD Scores in Curated Mismatch Repair Gene Variants Yields a Model for Clinical Validation and Prioritization. Human Mutation, 2015, 36, 712-719.	1.1	39
55	Novel pathogenic variants and genes for myopathies identified by whole exome sequencing. Molecular Genetics & Genomic Medicine, 2015, 3, 283-301.	0.6	43

#	Article	IF	Citations
56	Quantifying the heritability of glioma using genome-wide complex trait analysis. Scientific Reports, 2015, 5, 17267.	1.6	37
57	Identification of candidate regulatory SNPs by integrative analysis for prostate cancer genome data. , 2015, , .		1
58	Meta-analysis of genome-wide association studies identifies common susceptibility polymorphisms for colorectal and endometrial cancer near SH2B3 and TSHZ1. Scientific Reports, 2015, 5, 17369.	1.6	35
59	NAMPT and NAPRT1: novel polymorphisms and distribution of variants between normal tissues and tumor samples. Scientific Reports, 2014, 4, 6311.	1.6	21
60	Smoking Gun or Circumstantial Evidence? Comparison of Statistical Learning Methods using Functional Annotations for Prioritizing Risk Variants. Scientific Reports, 2015, 5, 13373.	1.6	7
61	An NGS Workflow Blueprint for DNA Sequencing Data and Its Application in Individualized Molecular Oncology. Cancer Informatics, 2015, 14s5, CIN.S30793.	0.9	10
62	Prioritization Of Nonsynonymous Single Nucleotide Variants For Exome Sequencing Studies Via Integrative Learning On Multiple Genomic Data. Scientific Reports, 2015, 5, 14955.	1.6	10
63	A Statistical Framework to Predict Functional Non-Coding Regions in the Human Genome Through Integrated Analysis of Annotation Data. Scientific Reports, 2015, 5, 10576.	1.6	144
64	Genome-wide Polygenic Burden of Rare Deleterious Variants in Sudden Unexpected Death in Epilepsy. EBioMedicine, 2015, 2, 1063-1070.	2.7	74
65	Disease-associated variants in different categories of disease located in distinct regulatory elements. BMC Genomics, 2015, 16, S3.	1.2	41
66	Computational methods and resources for the interpretation of genomic variants in cancer. BMC Genomics, 2015, 16, S7.	1.2	18
67	Rapid-Onset Obesity with Hypothalamic Dysfunction, Hypoventilation, and Autonomic Dysregulation (ROHHAD): exome sequencing of trios, monozygotic twins and tumours. Orphanet Journal of Rare Diseases, 2015, 10, 103.	1.2	45
68	Functional and Structural Consequence of Rare Exonic Single Nucleotide Polymorphisms: One Story, Two Tales. Genome Biology and Evolution, 2015, 7, 2929-2940.	1.1	12
69	Epilepsy with auditory features. Neurology: Genetics, 2015, 1, e5.	0.9	55
70	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. Human Mutation, 2015, 36, 1080-1087.	1.1	67
71	Concurrent DNA Copy-Number Alterations and Mutations in Genes Related to Maintenance of Genome Stability in Uninvolved Mammary Glandular Tissue from Breast Cancer Patients. Human Mutation, 2015, 36, 1088-1099.	1.1	11
72	Novel compound heterozygous mutations in PRKRA cause pure dystonia. Movement Disorders, 2015, 30, 877-878.	2.2	13
73	Novel CDKN2A mutations in Austrian melanoma patients. Melanoma Research, 2015, 25, 412-420.	0.6	10

#	Article	IF	Citations
74	ccKOPLS: Confounder-correcting kernel-based orthogonal projections to latent structures. , 2015, , .		1
75	HMMvar-func: a new method for predicting the functional outcome of genetic variants. BMC Bioinformatics, 2015, 16, 351.	1.2	10
76	Adult-onset autosomal dominant spastic paraplegia linked to a GTPase-effector domain mutation of dynamin 2. BMC Neurology, 2015, 15, 223.	0.8	39
77	Defects in fatty acid amide hydrolase 2 in a male with neurologic and psychiatric symptoms. Orphanet Journal of Rare Diseases, 2015, 10, 38.	1.2	19
78	The RAB39B p.G192R mutation causes X-linked dominant Parkinsonâ€™s disease. Molecular Neurodegeneration, 2015, 10, 50.	4.4	91
79	Microdeletions in 9q33.3-q34.11 in five patients with intellectual disability, microcephaly, and seizures of incomplete penetrance: is STXBP1 not the only causative gene?. Molecular Cytogenetics, 2015, 8, 72.	0.4	14
80	Phenotype-driven strategies for exome prioritization of human Mendelian disease genes. Genome Medicine, 2015, 7, 81.	3.6	97
81	Developments in our understanding of the genetic basis of birth defects. Birth Defects Research Part A: Clinical and Molecular Teratology, 2015, 103, 680-691.	1.6	30
82	Introduction to the analysis of next generation sequencing data and its application to venous thromboembolism. Thrombosis and Haemostasis, 2015, 114, 920-932.	1.8	10
83	Next-generation Sequencing of RYR1 and CACNA1S in Malignant Hyperthermia and Exertional Heat Illness. Anesthesiology, 2015, 122, 1033-1046.	1.3	70
84	KIAA0586is Mutated in Joubert Syndrome. Human Mutation, 2015, 36, 831-835.	1.1	62
85	Classification of Amino Acid Substitutions in Mismatch Repair Proteins Using PON-MMR2. Human Mutation, 2015, 36, 1128-1134.	1.1	17
86	Genome-wide variant analysis of simplex autism families with an integrative clinical-bioinformatics pipeline. Journal of Physical Education and Sports Management, 2015, 1, a000422.	0.5	6
87	Analysis of genetic variation and potential applications in genome-scale metabolic modeling. Frontiers in Bioengineering and Biotechnology, 2015, 3, 13.	2.0	30
88	Integrative analysis of functional genomic annotations and sequencing data to identify rare causal variants via hierarchical modeling. Frontiers in Genetics, 2015, 6, 17.	1.1	2
89	Anatomy of protein disorder, flexibility and disease-related mutations. Frontiers in Molecular Biosciences, 2015, 2, 47.	1.6	16
90	A De Novo Mutation inTEAD1Causes Nonâ€“X-Linked Aicardi Syndrome. , 2015, 56, 3896.		22
91	Whole Genome Sequencing Reveals a De Novo SHANK3 Mutation in Familial Autism Spectrum Disorder. PLoS ONE, 2015, 10, e0116358.	1.1	55

#	Article	IF	Citations
92	Insight into Neutral and Disease-Associated Human Genetic Variants through Interpretable Predictors. PLoS ONE, 2015, 10, e0120729.	1.1	2
93	Lessons Learned from Whole Exome Sequencing in Multiplex Families Affected by a Complex Genetic Disorder, Intracranial Aneurysm. PLoS ONE, 2015, 10, e0121104.	1.1	32
94	A Dual Model for Prioritizing Cancer Mutations in the Non-coding Genome Based on Germline and Somatic Events. PLoS Computational Biology, 2015, 11, e1004583.	1.5	17
95	The Intolerance of Regulatory Sequence to Genetic Variation Predicts Gene Dosage Sensitivity. PLoS Genetics, 2015, 11, e1005492.	1.5	123
96	Sequence to Medical Phenotypes: A Framework for Interpretation of Human Whole Genome DNA Sequence Data. PLoS Genetics, 2015, 11, e1005496.	1.5	23
97	Whole Exome Sequencing Reveals Novel PHEX Splice Site Mutations in Patients with Hypophosphatemic Rickets. PLoS ONE, 2015, 10, e0130729.	1.1	32
98	A Frame-Shift Mutation in CAV1 Is Associated with a Severe Neonatal Progeroid and Lipodystrophy Syndrome. PLoS ONE, 2015, 10, e0131797.	1.1	46
99	Two Novel De Novo GARS Mutations Cause Early-Onset Axonal Charcot-Marie-Tooth Disease. PLoS ONE, 2015, 10, e0133423.	1.1	22
100	Expression Quantitative Trait Loci Information Improves Predictive Modeling of Disease Relevance of Non-Coding Genetic Variation. PLoS ONE, 2015, 10, e0140758.	1.1	17
101	Mutation Screening of 1,237 Cancer Genes across Six Model Cell Lines of Basal-Like Breast Cancer. PLoS ONE, 2015, 10, e0144528.	1.1	6
102	A Single Amino Acid Deletion (Î”F1502) in the S6 Segment of CaV2.1 Domain III Associated with Congenital Ataxia Increases Channel Activity and Promotes Ca2+ Influx. PLoS ONE, 2015, 10, e0146035.	1.1	22
103	Identification of deep intronic variants in 15 haemophilia A patients by next generation sequencing of the whole factor VIII gene. Thrombosis and Haemostasis, 2015, 114, 757-767.	1.8	31
104	Identifying Highly Penetrant Disease Causal Mutations Using Next Generation Sequencing: Guide to Whole Process. BioMed Research International, 2015, 2015, 1-16.	0.9	7
105	Functional genome-wide siRNA screen identifies KIAA0586 as mutated in Joubert syndrome. ELife, 2015, 4, e06602.	2.8	64
106	Human Enhancers Are Fragile and Prone to Deactivating Mutations. Molecular Biology and Evolution, 2015, 32, 2161-2180.	3.5	17
107	Exome sequencing and array-based comparative genomic hybridisation analysis of preferential 6-methylmercaptopurine producers. Pharmacogenomics Journal, 2015, 15, 414-421.	0.9	5
108	SDHB/SDHA immunohistochemistry in pheochromocytomas and paragangliomas: a multicenter interobserver variation analysis using virtual microscopy: a Multinational Study of the European Network for the Study of Adrenal Tumors (ENS@T). Modern Pathology, 2015, 28, 807-821.	2.9	176
110	Human HOIP and LUBAC deficiency underlies autoinflammation, immunodeficiency, amylopectinosis, and lymphangiectasia. Journal of Experimental Medicine, 2015, 212, 939-951.	4.2	241

#	Article	IF	Citations
111	Recessive Mutations in the Î±3 (VI) Collagen Gene COL6A3 Cause Early-Onset Isolated Dystonia. American Journal of Human Genetics, 2015, 96, 883-893.	2.6	79
112	A Missense Mutation in KCTD17 Causes Autosomal Dominant Myoclonus-Dystonia. American Journal of Human Genetics, 2015, 96, 938-947.	2.6	109
113	Novel, Compound Heterozygous, Single-Nucleotide Variants inMARS2Associated with Developmental Delay, Poor Growth, and Sensorineural Hearing Loss. Human Mutation, 2015, 36, 587-592.	1.1	29
114	Juvenile myelomonocytic leukemia due to a germline CBL Y371C mutation: 35-year follow-up of a large family. Human Genetics, 2015, 134, 775-787.	1.8	21
115	Individualized Iterative Phenotyping for Genome-wide Analysis of Loss-of-Function Mutations. American Journal of Human Genetics, 2015, 96, 913-925.	2.6	66
116	Genetic diagnosis of developmental disorders in the DDD study: a scalable analysis of genome-wide research data. Lancet, The, 2015, 385, 1305-1314.	6.3	651
117	Cerebral palsy: causes, pathways, and the role of genetic variants. American Journal of Obstetrics and Gynecology, 2015, 213, 779-788.	0.7	290
118	Sequential data selection for predicting the pathogenic effects of sequence variation. , 2015, , .		1
119	A positive genotypeâ€“phenotype correlation in a large cohort of patients with Pseudohypoparathyroidism Type Ia and Pseudoâ€pseudohypoparathyroidism and 33 newly identified mutations in the <scp>GNAS</scp> gene. Molecular Genetics & Genomic Medicine, 2015, 3, 111-120.	0.6	46
120	Targeted Nextâ€Generation Sequencing Analysis of 1,000 Individuals with Intellectual Disability. Human Mutation, 2015, 36, 1197-1204.	1.1	161
121	Genomeâ€wide association study of neocortical Lewyâ€related pathology. Annals of Clinical and Translational Neurology, 2015, 2, 920-931.	1.7	25
122	Network-Informed Gene Ranking Tackles Genetic Heterogeneity in Exome-Sequencing Studies of Monogenic Disease. Human Mutation, 2015, 36, 1135-1144.	1.1	7
123	Rare inactivating PDE11A variants associated with testicular germ cell tumors. Endocrine-Related Cancer, 2015, 22, 909-917.	1.6	24
124	Comparison of predicted and actual consequences of missense mutations. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, E5189-98.	3.3	200
125	Homozygous loss-of-function variants in European cosmopolitan and isolate populations. Human Molecular Genetics, 2015, 24, 5464-5474.	1.4	27
126	BayesPI-BAR: a new biophysical model for characterization of regulatory sequence variations. Nucleic Acids Research, 2015, 43, gkv733.	6.5	17
127	Autosomal-Recessive Intellectual Disability with Cerebellar Atrophy Syndrome Caused by Mutation of the Manganese and Zinc Transporter Gene SLC39A8. American Journal of Human Genetics, 2015, 97, 886-893.	2.6	171
128	Sixteen new lung function signals identified through 1000 Genomes Project reference panel imputation. Nature Communications, 2015, 6, 8658.	5.8	108

#	Article	IF	Citations
129	Contribution of Rare and Common Genetic Variants to Plasma Lipid Levels and Carotid Stiffness and Geometry. Circulation: Cardiovascular Genetics, 2015, 8, 628-636.	5.1	21
130	A Note on the Evaluation of Mutation Prioritization Algorithms. , 2015, , .		0
131	Apparent underdiagnosis of Cerebrotendinous Xanthomatosis revealed by analysis of ~60,000 human exomes. Molecular Genetics and Metabolism, 2015, 116, 298-304.	0.5	79
132	A de novo whole gene deletion of XIAP detected by exome sequencing analysis in very early onset inflammatory bowel disease: a case report. BMC Gastroenterology, 2015, 15, 160.	0.8	38
133	A novel Fanconi anaemia subtype associated with a dominant-negative mutation in RAD51. Nature Communications, 2015, 6, 8829.	5.8	130
134	Comparison and integration of deleteriousness prediction methods for nonsynonymous SNVs in whole exome sequencing studies. Human Molecular Genetics, 2015, 24, 2125-2137.	1.4	892
135	Targeted next-generation sequencing panels for monogenetic disorders in clinical diagnostics: the opportunities and challenges. Expert Review of Molecular Diagnostics, 2015, 15, 61-70.	1.5	44
136	Use of Whole Genome Sequencing for Diagnosis and Discovery in the Cancer Genetics Clinic. EBioMedicine, 2015, 2, 74-81.	2.7	50
137	Role of common and rare variants in SCN10A: results from the Brugada syndrome QRS locus gene discovery collaborative study. Cardiovascular Research, 2015, 106, 520-529.	1.8	108
138	Whole-exome sequencing points to considerable genetic heterogeneity of cerebral palsy. Molecular Psychiatry, 2015, 20, 176-182.	4.1	178
139	SeqHBase: a big data toolset for family based sequencing data analysis. Journal of Medical Genetics, 2015, 52, 282-288.	1.5	17
140	Identification of altered cis-regulatory elements in human disease. Trends in Genetics, 2015, 31, 67-76.	2.9	99
141	Whole-genome sequencing of quartet families with autism spectrum disorder. Nature Medicine, 2015, 21, 185-191.	15.2	457
142	Whole-exome sequencing in undiagnosed genetic diseases: interpreting 119 trios. Genetics in Medicine, 2015, 17, 774-781.	1.1	284
143	Resolving clinical diagnoses for syndromic cleft lip and/or palate phenotypes using wholeâ€exome sequencing. Clinical Genetics, 2015, 88, 441-449.	1.0	14
144	Using the plurality of codon positions to identify deleterious variants in human exomes. Bioinformatics, 2015, 31, 301-305.	1.8	3
145	MAT2A Mutations Predispose Individuals to Thoracic Aortic Aneurysms. American Journal of Human Genetics, 2015, 96, 170-177.	2.6	92
146	Exome sequencing in HFE C282Y homozygous men with extreme phenotypes identifies a GNPAT variant associated with severe iron overload. Hepatology, 2015, 62, 429-439.	3.6	75

#	Article	IF	Citations
147	DDIG-in: detecting disease-causing genetic variations due to frameshifting indels and nonsense mutations employing sequence and structural properties at nucleotide and protein levels. Bioinformatics, 2015, 31, 1599-1606.	1.8	52
148	A method for calculating probabilities of fitness consequences for point mutations across the human genome. Nature Genetics, 2015, 47, 276-283.	9.4	247
149	Breast Cancer Genomics From Microarrays to Massively Parallel Sequencing: Paradigms and New Insights. Journal of the National Cancer Institute, 2015, 107, .	3.0	80
150	New Tools for Mendelian Disease Gene Identification: PhenoDB Variant Analysis Module; and GeneMatcher, a Web-Based Tool for Linking Investigators with an Interest in the Same Gene. Human Mutation, 2015, 36, 425-431.	1.1	141
151	Genome-wide association analysis of more than 120,000 individuals identifies 15 new susceptibility loci for breast cancer. Nature Genetics, 2015, 47, 373-380.	9.4	513
152	Determining the prevalence of McArdle disease from gene frequency by analysis of next-generation sequencing data. Genetics in Medicine, 2015, 17, 1002-1006.	1.1	46
153	Homozygous SLC6A17 Mutations Cause Autosomal-Recessive Intellectual Disability with Progressive Tremor, Speech Impairment, and Behavioral Problems. American Journal of Human Genetics, 2015, 96, 386-396.	2.6	27
154	Prioritizing genes for X-linked diseases using population exome data. Human Molecular Genetics, 2015, 24, 599-608.	1.4	18
155	Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genetics in Medicine, 2015, 17, 405-424.	1.1	20,455
156	The Evaluation of Tools Used to Predict the Impact of Missense Variants Is Hindered by Two Types of Circularity. Human Mutation, 2015, 36, 513-523.	1.1	283
157	Mutations in DYNC2LI1 disrupt cilia function and cause short rib polydactyly syndrome. Nature Communications, 2015, 6, 7092.	5.8	79
158	A method to predict the impact of regulatory variants from DNA sequence. Nature Genetics, 2015, 47, 955-961.	9.4	416
159	CHD2variants are a risk factor for photosensitivity in epilepsy. Brain, 2015, 138, 1198-1208.	3.7	112
160	Homozygous mutation of STXBP5L explains an autosomal recessive infantile-onset neurodegenerative disorder. Human Molecular Genetics, 2015, 24, 2000-2010.	1.4	25
161	Running spell-check to identify regulatory variants. Nature Genetics, 2015, 47, 853-855.	9.4	5
162	Resequencing and association analysis of coding regions at twenty candidate genes suggest a role for rare risk variation at AKAP9 and protective variation at NRXN1 in schizophrenia susceptibility. Journal of Psychiatric Research, 2015, 66-67, 38-44.	1.5	18
163	Identical by descent L1CAM mutation in two apparently unrelated families with intellectual disability without L1 syndrome. European Journal of Medical Genetics, 2015, 58, 364-368.	0.7	17
164	Targeted Sequencing in Chromosome 17q Linkage Region Identifies Familial Glioma Candidates in the Gliogene Consortium. Scientific Reports, 2015, 5, 8278.	1.6	22

#	Article	IF	Citations
165	Predicting the sequence specificities of DNA- and RNA-binding proteins by deep learning. Nature Biotechnology, 2015, 33, 831-838.	9.4	2,206
166	Incorporating Functional Information in Tests of Excess De Novo Mutational Load. American Journal of Human Genetics, 2015, 97, 272-283.	2.6	29
167	An inÂvivo genetic reversion highlights the crucial role of Myb-Like, SWIRM, and MPN domains 1 (MYSM1) in human hematopoiesis and lymphocyte differentiation. Journal of Allergy and Clinical Immunology, 2015, 136, 1619-1626.e5.	1.5	63
168	Missense mutations inTENM4, a regulator of axon guidance and central myelination, cause essential tremor. Human Molecular Genetics, 2015, 24, 5677-5686.	1.4	134
169	The genetics of human infertility by functional interrogation of SNPs in mice. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 10431-10436.	3.3	62
170	Functional Characterization of Rare Variants Implicated in Susceptibility to Lone Atrial Fibrillation. Circulation: Arrhythmia and Electrophysiology, 2015, 8, 1095-1104.	2.1	44
171	Non-coding genetic variants in human disease: Figure 1.. Human Molecular Genetics, 2015, 24, R102-R110.	1.4	466
172	Impairment of immunity to Candida and Mycobacterium in humans with bi-allelic RORC mutations. Science, 2015, 349, 606-613.	6.0	366
173	Rare A2ML1 variants confer susceptibility to otitis media. Nature Genetics, 2015, 47, 917-920.	9.4	38
174	Mutations inGMPPBcause congenital myasthenic syndrome and bridge myasthenic disorders with dystroglycanopathies. Brain, 2015, 138, 2493-2504.	3.7	111
175	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215.	2.6	574
176	Inherited coding variants at the CDKN2A locus influence susceptibility to acute lymphoblastic leukaemia in children. Nature Communications, 2015, 6, 7553.	5.8	72
177	Evolutionary Constraint and Disease Associations of Post-Translational Modification Sites in Human Genomes. PLoS Genetics, 2015, 11, e1004919.	1.5	69
178	Genes and Podocytes Ã¢â‚¬â€œ New Insights into Mechanisms of Podocytopathy. Frontiers in Endocrinology, 2014, 5, 226.	1.5	52
179	A new GWAS and meta-analysis with 1000Genomes imputation identifies novel risk variants for colorectal cancer. Scientific Reports, 2015, 5, 10442.	1.6	109
180	Lysoplex: An efficient toolkit to detect DNA sequence variations in the autophagy-lysosomal pathway. Autophagy, 2015, 11, 928-938.	4.3	47
181	Joubert syndrome: a model for untangling recessive disorders with extreme genetic heterogeneity. Journal of Medical Genetics, 2015, 52, 514-522.	1.5	219
182	Pulmonary Nontuberculous Mycobacterial Infection. A Multisystem, Multigenic Disease. American Journal of Respiratory and Critical Care Medicine, 2015, 192, 618-628.	2.5	136

#	Article	IF	Citations
183	Massively Parallel Functional Analysis of BRCA1 RING Domain Variants. Genetics, 2015, 200, 413-422.	1.2	272
184	Exome Sequencing Analysis Reveals Variants in Primary Immunodeficiency Genes in Patients With Very Early Onset Inflammatory Bowel Disease. Gastroenterology, 2015, 149, 1415-1424.	0.6	99
185	Activating CARD14 Mutations Are Associated with Generalized Pustular Psoriasis but Rarely Account for Familial Recurrence in Psoriasis Vulgaris. Journal of Investigative Dermatology, 2015, 135, 2964-2970.	0.3	89
186	Recognizable cerebellar dysplasia associated with mutations in multiple tubulin genes. Human Molecular Genetics, 2015, 24, 5313-5325.	1.4	77
187	Mutations in the genes encoding eukaryotic translation initiation factor 2B in Japanese patients with vanishing white matter disease. Brain and Development, 2015, 37, 960-966.	0.6	10
188	HCFC1 loss-of-function mutations disrupt neuronal and neural progenitor cells of the developing brain. Human Molecular Genetics, 2015, 24, 3335-3347.	1.4	47
189	Adult-onset painful axonal polyneuropathy caused by a dominantNAGLUmutation. Brain, 2015, 138, 1477-1483.	3.7	24
190	Whole exome sequencing combined with integrated variant annotation prediction identifies asymptomatic Tangier disease with compound heterozygous mutations in ABCA1 gene. Atherosclerosis, 2015, 240, 324-329.	0.4	16
191	Re-annotation of presumed noncoding disease/trait-associated genetic variants by integrative analyses. Scientific Reports, 2015, 5, 9453.	1.6	13
192	Mutational analysis of MATR3 in Taiwanese patients with amyotrophic lateral sclerosis. Neurobiology of Aging, 2015, 36, 2005.e1-2005.e4.	1.5	64
193	Mutations Impairing GSK3-Mediated MAF Phosphorylation Cause Cataract, Deafness, Intellectual Disability, Seizures, and a Down Syndrome-like Facies. American Journal of Human Genetics, 2015, 96, 816-825.	2.6	102
194	The genotypic and phenotypic spectrum of PIGA deficiency. Orphanet Journal of Rare Diseases, 2015, 10, 23.	1.2	70
195	Rare variant association studies: considerations, challenges and opportunities. Genome Medicine, 2015, 7, 16.	3.6	176
196	Connecting the CNTNAP2 Networks with Neurodevelopmental Disorders. Molecular Syndromology, 2015, 6, 7-22.	0.3	89
197	Utilization of Multigene Panels in Hereditary Cancer Predisposition Testing. , 2015, , 459-482.		0
198	Meta-analysis of the TNFAIP3 region in psoriasis reveals a risk haplotype that is distinct from other autoimmune diseases. Genes and Immunity, 2015, 16, 120-126.	2.2	29
199	An integrative approach to predicting the functional effects of non-coding and coding sequence variation. Bioinformatics, 2015, 31, 1536-1543.	1.8	524
200	Longevity Genes. Advances in Experimental Medicine and Biology, 2015, , .	0.8	5

#	Article	IF	Citations
201	Exome and Whole Genome Sequencing in Aging and Longevity. Advances in Experimental Medicine and Biology, 2015, 847, 127-139.	0.8	5
202	Types and effects of protein variations. Human Genetics, 2015, 134, 405-421.	1.8	29
203	Life-threatening influenza and impaired interferon amplification in human IRF7 deficiency. Science, 2015, 348, 448-453.	6.0	389
204	Rare variants in neuronal excitability genes influence risk for bipolar disorder. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 3576-3581.	3.3	152
205	Identifying causal regulatory SNPs in ChIP-seq enhancers. Nucleic Acids Research, 2015, 43, 225-236.	6.5	50
206	Î±IIbÎ²3 variants defined by next-generation sequencing: Predicting variants likely to cause Glanzmann thrombasthenia. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, E1898-907.	3.3	36
207	Next Generation Sequencing in Cancer Research, Volume 2. , 2015, , .		4
208	Scripps Genome ADVISER: Annotation and Distributed Variant Interpretation SERver. PLoS ONE, 2015, 10, e0116815.	1.1	17
209	Excess of rare, inherited truncating mutations in autism. Nature Genetics, 2015, 47, 582-588.	9.4	531
210	A review of study designs and statistical methods for genomic epidemiology studies using next generation sequencing. Frontiers in Genetics, 2015, 6, 149.	1.1	48
211	Machine learning applications in genetics and genomics. Nature Reviews Genetics, 2015, 16, 321-332.	7.7	1,334
212	Mutations in KCNH1 and ATP6V1B2 cause Zimmermann-Laband syndrome. Nature Genetics, 2015, 47, 661-667.	9.4	177
213	Effect of predicted protein-truncating genetic variants on the human transcriptome. Science, 2015, 348, 666-669.	6.0	252
214	Autosomal-Dominant Multiple Pterygium Syndrome Is Caused by Mutations in MYH3. American Journal of Human Genetics, 2015, 96, 841-849.	2.6	55
215	Identifying functional noncoding variants from genome-wide association studies for cardiovascular disease and related traits. Current Opinion in Lipidology, 2015, 26, 120-126.	1.2	7
216	Next-generation gene discovery for variants of large impact on lipid traits. Current Opinion in Lipidology, 2015, 26, 114-119.	1.2	5
217	Whole-genome sequencing is more powerful than whole-exome sequencing for detecting exome variants. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 5473-5478.	3.3	475
218	Translational Research Methods: Basics of Renal Molecular Biology. , 2015, , 1-22.		0

#	Article	IF	Citations
219	Truncated HSPB1 causes axonal neuropathy and impairs tolerance to unfolded protein stress. BBA Clinical, 2015, 3, 233-242.	4.1	26
220	Non-syndromic retinitis pigmentosa due to mutations in the mucopolysaccharidosis type IIIC gene, heparan-alpha-glucosaminide N-acetyltransferase (HGSNAT). Human Molecular Genetics, 2015, 24, 3742-51.	1.4	47
221	Genomic approaches for understanding the genetics of complex disease. Genome Research, 2015, 25, 1432-1441.	2.4	75
222	Genome-wide association study identifies multiple susceptibility loci for glioma. Nature Communications, 2015, 6, 8559.	5.8	112
223	The origins, determinants, and consequences of human mutations. Science, 2015, 349, 1478-1483.	6.0	143
224	Somatic mutation in cancer and normal cells. Science, 2015, 349, 1483-1489.	6.0	996
225	Diving deeper to predict noncoding sequence function. Nature Methods, 2015, 12, 925-926.	9.0	4
226	Successful diagnosis of HIBCH deficiency from exome sequencing and positive retrospective analysis of newborn screening cards in two siblings presenting with Leigh's disease. Molecular Genetics and Metabolism, 2015, 115, 161-167.	0.5	32
227	Parlez-vous VUS?. Genome Research, 2015, 25, 1423-1426.	2.4	36
228	Compound heterozygous mutations in the noncoding RNU4ATAC cause Roifman Syndrome by disrupting minor intron splicing. Nature Communications, 2015, 6, 8718.	5.8	104
229	Prospective participant selection and ranking to maximize actionable pharmacogenetic variants and discovery in the eMERGE Network. Genome Medicine, 2015, 7, 67.	3.6	23
230	Germline genetic variation in ETV6 and risk of childhood acute lymphoblastic leukaemia: a systematic genetic study. Lancet Oncology, The, 2015, 16, 1659-1666.	5.1	161
231	Assessing models for genetic prediction of complex traits: a comparison of visualization and quantitative methods. BMC Genomics, 2015, 16, 405.	1.2	0
232	Germline Heterozygous Variants in SEC23B Are Associated with Cowden Syndrome and Enriched in Apparently Sporadic Thyroid Cancer. American Journal of Human Genetics, 2015, 97, 661-676.	2.6	76
233	Large-scale identification of sequence variants influencing human transcription factor occupancy in vivo. Nature Genetics, 2015, 47, 1393-1401.	9.4	202
234	The human gene damage index as a gene-level approach to prioritizing exome variants. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 13615-13620.	3.3	213
235	Mutations in the TLR3 signaling pathway and beyond in adult patients with herpes simplex encephalitis. Genes and Immunity, 2015, 16, 552-566.	2.2	75
236	Expression quantitative trait locus analysis for translational medicine. Genome Medicine, 2015, 7, 60.	3.6	69

#	Article	IF	Citations
237	Genome-wide significant association with seven novel multiple sclerosis risk loci. Journal of Medical Genetics, 2015, 52, 848-855.	1.5	34
238	Mining the coding and non-coding genome for cancer drivers. Cancer Letters, 2015, 369, 307-315.	3.2	15
239	Building the foundation for genomics in precision medicine. Nature, 2015, 526, 336-342.	13.7	376
240	OncoRep: an n-of-1 reporting tool to support genome-guided treatment for breast cancer patients using RNA-sequencing. BMC Medical Genomics, 2015, 8, 24.	0.7	12
241	Rare variant discovery by deep whole-genome sequencing of 1,070 Japanese individuals. Nature Communications, 2015, 6, 8018.	5.8	352
242	Predicting effects of noncoding variants with deep learningâ€“based sequence model. Nature Methods, 2015, 12, 931-934.	9.0	1,714
243	New basal cell carcinoma susceptibility loci. Nature Communications, 2015, 6, 6825.	5.8	59
244	Functional Analysis of a Carotid Intima-Media Thickness Locus Implicates BCAR1 and Suggests a Causal Variant. Circulation: Cardiovascular Genetics, 2015, 8, 696-706.	5.1	17
245	Targeted combination nanotherapeutics in cancer a real promise. Nanomedicine, 2015, 10, 1855-1857.	1.7	2
246	Prediction of Causal Candidate Genes in Coronary Artery Disease Loci. Arteriosclerosis, Thrombosis, and Vascular Biology, 2015, 35, 2207-2217.	1.1	101
247	Prevalence of PALB2 mutations in Australian familial breast cancer cases and controls. Breast Cancer Research, 2015, 17, 111.	2.2	36
248	Can the impact of human genetic variations be predicted?. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 11426-11427.	3.3	26
249	Systematic Validation of RNF213 Coding Variants in Japanese Patients With Moyamoya Disease. Journal of the American Heart Association, 2015, 4, .	1.6	59
250	Genome-wide association analyses based on whole-genome sequencing in Sardinia provide insights into regulation of hemoglobin levels. Nature Genetics, 2015, 47, 1264-1271.	9.4	66
251	Genome sequencing elucidates Sardinian genetic architecture and augments association analyses for lipid and blood inflammatory markers. Nature Genetics, 2015, 47, 1272-1281.	9.4	193
252	Computational approaches to study the effects of small genomic variations. Journal of Molecular Modeling, 2015, 21, 251.	0.8	21
253	Modeling Human Severe Combined Immunodeficiency and Correction by CRISPR/Cas9-Enhanced Gene Targeting. Cell Reports, 2015, 12, 1668-1677.	2.9	95
254	Actionable exomic incidental findings in 6503 participants: challenges of variant classification. Genome Research, 2015, 25, 305-315.	2.4	313

#	Article	IF	Citations
255	Whole exome sequencing identifies a novel frameshift mutation in GPC3 gene in a patient with overgrowth syndrome. Gene, 2015, 572, 303-306.	1.0	16
256	Whole-genome sequencing suggests a chemokine gene cluster that modifies age at onset in familial Alzheimer's disease. Molecular Psychiatry, 2015, 20, 1294-1300.	4.1	55
257	Genomic variant annotation and prioritization with ANNOVAR and wANNOVAR. Nature Protocols, 2015, 10, 1556-1566.	5.5	727
258	Greenlandic Inuit show genetic signatures of diet and climate adaptation. Science, 2015, 349, 1343-1347.	6.0	397
259	Genome-wide Association Analysis of Psoriatic Arthritis and Cutaneous Psoriasis Reveals Differences in Their Genetic Architecture. American Journal of Human Genetics, 2015, 97, 816-836.	2.6	245
260	Exome and regulatory element sequencing of neuromyelitis optica patients. Journal of Neuroimmunology, 2015, 289, 139-142.	1.1	6
261	The evolving spectrum ofPRRT2-associated paroxysmal diseases. Brain, 2015, 138, 3476-3495.	3.7	218
262	Histone Modifier Genes Alter Conotruncal Heart Phenotypes in 22q11.2 Deletion Syndrome. American Journal of Human Genetics, 2015, 97, 869-877.	2.6	49
263	An ensemble approach to accurately detect somatic mutations using SomaticSeq. Genome Biology, 2015, 16, 197.	3.8	93
264	The role of functional data in interpreting the effects of genetic variation. Molecular Biology of the Cell, 2015, 26, 3904-3908.	0.9	13
265	Toward an improved definition of the genetic and tumor spectrum associated with SDH germ-line mutations. Genetics in Medicine, 2015, 17, 610-620.	1.1	91
266	The human splicing code reveals new insights into the genetic determinants of disease. Science, 2015, 347, 1254806.	6.0	1,053
267	Genetic testing for RAD51C mutations: in the clinic and community. Clinical Genetics, 2015, 88, 303-312.	1.0	39
268	Bioinformatics for Clinical Next Generation Sequencing. Clinical Chemistry, 2015, 61, 124-135.	1.5	114
269	The Mutational Landscape in Pediatric Acute Lymphoblastic Leukemia Deciphered by Whole Genome Sequencing. Human Mutation, 2015, 36, 118-128.	1.1	38
270	The discovery of integrated gene networks for autism and related disorders. Genome Research, 2015, 25, 142-154.	2.4	259
271	A recurrent de novo mutation in KCNC1 causes progressive myoclonus epilepsy. Nature Genetics, 2015, 47, 39-46.	9.4	245
272	Ensembl 2015. Nucleic Acids Research, 2015, 43, D662-D669.	6.5	1,145

#	Article	IF	Citations
273	Recent genetic findings in schizophrenia and their therapeutic relevance. Journal of Psychopharmacology, 2015, 29, 85-96.	2.0	157
274	DANN: a deep learning approach for annotating the pathogenicity of genetic variants. Bioinformatics, 2015, 31, 761-763.	1.8	786
275	Exploring the function of genetic variants in the non-coding genomic regions: approaches for identifying human regulatory variants affecting gene expression. Briefings in Bioinformatics, 2015, 16, 393-412.	3.2	58
276	Current trend of annotating single nucleotide variation in humans â€“ A case study on SNVrap. Methods, 2015, 79-80, 32-40.	1.9	12
277	Targeted Genes Sequencing Identified a Novel 15 bp Deletion on GJA8 in a Chinese Family with Autosomal Dominant Congenital Cataracts. Chinese Medical Journal, 2016, 129, 860-867.	0.9	8
279	Extended clinical and genetic spectrum associated with biallelic RTEL1 mutations. Blood Advances, 2016, 1, 36-46.	2.5	19
280	Challenges ahead for matchmaking. IT - Information Technology, 2016, 58, 140-144.	0.6	0
281	Authors' Response to Wong et al.. Genetic Testing and Molecular Biomarkers, 2016, 20, 414-415.	0.3	0
282	Prevalence of BRCA1/BRCA2 mutations in a Brazilian population sample at-risk for hereditary breast cancer and characterization of its genetic ancestry. Oncotarget, 2016, 7, 80465-80481.	0.8	62
283	Setting Up a Laboratory. , 2016, , 409-426.		1
284	Identification of Susceptibility Genes of Adult Asthma in French Canadian Women. Canadian Respiratory Journal, 2016, 2016, 1-12.	0.8	10
285	A Survey of Computational Tools to Analyze and Interpret Whole Exome Sequencing Data. International Journal of Genomics, 2016, 2016, 1-16.	0.8	37
286	Biallelic Mutations inCRB1Underlie Autosomal Recessive Familial Foveal Retinoschisis. , 2016, 57, 2637.		34
287	Quantifying deleterious effects of regulatory variants. Nucleic Acids Research, 2016, 45, gkw1263.	6.5	14
288	Fine-Mapping of 18q21.1 Locus Identifies Single Nucleotide Polymorphisms Associated with Nonsyndromic Cleft Lip with or without Cleft Palate. Frontiers in Genetics, 2016, 7, 88.	1.1	3
289	Genetic Diagnosis Using Whole Exome Sequencing in Common Variable Immunodeficiency. Frontiers in Immunology, 2016, 7, 220.	2.2	247
290	A Hypothesis for Using Pathway Genetic Load Analysis for Understanding Complex Outcomes in Bilirubin Encephalopathy. Frontiers in Neuroscience, 2016, 10, 376.	1.4	14
291	The role of small in-frame insertions/deletions in inherited eye disorders and how structural modelling can help estimate their pathogenicity. Orphanet Journal of Rare Diseases, 2016, 11, 125.	1.2	13

#	Article	IF	Citations
292	Genetic Evidence for Elevated Pathogenicity of Mitochondrial DNA Heteroplasmy in Autism Spectrum Disorder. PLoS Genetics, 2016, 12, e1006391.	1.5	52
293	DNAH6 and Its Interactions with PCD Genes in Heterotaxy and Primary Ciliary Dyskinesia. PLoS Genetics, 2016, 12, e1005821.	1.5	92
294	Quantitative Trait Loci Identify Functional Noncoding Variation in Cancer. PLoS Genetics, 2016, 12, e1005826.	1.5	7
295	Identification and Functional Testing of ERCC2 Mutations in a Multi-national Cohort of Patients with Familial Breast- and Ovarian Cancer. PLoS Genetics, 2016, 12, e1006248.	1.5	22
296	Family Based Whole Exome Sequencing Reveals the Multifaceted Role of Notch Signaling in Congenital Heart Disease. PLoS Genetics, 2016, 12, e1006335.	1.5	59
297	A Large Genome-Wide Association Study of Age-Related Hearing Impairment Using Electronic Health Records. PLoS Genetics, 2016, 12, e1006371.	1.5	78
298	Genome Sequence Variability Predicts Drug Precautions and Withdrawals from the Market. PLoS ONE, 2016, 11, e0162135.	1.1	17
299	Genomic Profile of Chronic Lymphocytic Leukemia in Korea Identified by Targeted Sequencing. PLoS ONE, 2016, 11, e0167641.	1.1	27
300	Systems psychopharmacology: A network approach to developing novel therapies. World Journal of Psychiatry, 2016, 6, 66.	1.3	15
301	Novel ADAMTSL2-mutations in a patient with geleophysic dysplasia type I. Clinical Dysmorphology, 2016, 25, 106-109.	0.1	5
302	Rare Variants in Cardiomyopathy Genes Associated With Stress-Induced Cardiomyopathy. Neurosurgery, 2016, 78, 835-843.	0.6	3
303	Functional Analysis of Missense Variants in the Putative Breast Cancer Susceptibility GeneXRCC2. Human Mutation, 2016, 37, 914-925.	1.1	12
304	Ectrodactyly and Lethal Pulmonary Acinar Dysplasia Associated with HomozygousFGFR2Mutations Identified by Exome Sequencing. Human Mutation, 2016, 37, 955-963.	1.1	30
305	A Role for the Chromatinâ€Remodeling FactorBAZ1Ain Neurodevelopment. Human Mutation, 2016, 37, 964-975.	1.1	29
306	Recurrent mutations of the exportin 1 gene (XPO1) and their impact on selective inhibitor of nuclear export compounds sensitivity in primary mediastinal Bâ€cell lymphoma. American Journal of Hematology, 2016, 91, 923-930.	2.0	79
307	Biallelic CACNA1A mutations cause early onset epileptic encephalopathy with progressive cerebral, cerebellar, and optic nerve atrophy. American Journal of Medical Genetics, Part A, 2016, 170, 2173-2176.	0.7	65
308	CRIPT exonic deletion and a novel missense mutation in a female with short stature, dysmorphic features, microcephaly, and pigmentary abnormalities. American Journal of Medical Genetics, Part A, 2016, 170, 2206-2211.	0.7	16
309	Spondyloocular Syndrome: Novel Mutations in XYLT2 Gene and Expansion of the Phenotypic Spectrum. Journal of Bone and Mineral Research, 2016, 31, 1577-1585.	3.1	43

#	Article	IF	Citations
310	Functional annotation of noncoding variants and prioritization of cancer-associated lncRNAs in lung cancer. Oncology Letters, 2016, 12, 222-230.	0.8	8
311	Advances in the genetics of schizophrenia: toward a network and pathway view for drug discovery. Annals of the New York Academy of Sciences, 2016, 1366, 61-75.	1.8	14
312	Mutation inADORA1identified as likely cause of early-onset parkinsonism and cognitive dysfunction. Movement Disorders, 2016, 31, 1004-1011.	2.2	38
313	Clinical and genetic features of cervical dystonia in a large multicenter cohort. Neurology: Genetics, 2016, 2, e69.	0.9	44
314	Spontaneous shaker rat mutant â€“ a new model for X-linked tremor-ataxia. DMM Disease Models and Mechanisms, 2016, 9, 553-62.	1.2	17
315	Mutations inFLNCare Associated with Familial Restrictive Cardiomyopathy. Human Mutation, 2016, 37, 269-279.	1.1	138
316	Prioritizing Variants in Complete Hereditary Breast and Ovarian Cancer Genes in Patients Lacking KnownBRCAMutations. Human Mutation, 2016, 37, 640-652.	1.1	39
317	MSeqDR: A Centralized Knowledge Repository and Bioinformatics Web Resource to Facilitate Genomic Investigations in Mitochondrial Disease. Human Mutation, 2016, 37, 540-548.	1.1	42
318	Complex mode of inheritance in holoprosencephaly revealed by whole exome sequencing. Clinical Genetics, 2016, 89, 659-668.	1.0	36
319	ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. Human Mutation, 2016, 37, 653-660.	1.1	40
320	Variation Interpretation Predictors: Principles, Types, Performance, and Choice. Human Mutation, 2016, 37, 579-597.	1.1	109
321	Whole exome sequencing reveals a C-terminal germline variant in CEBPA-associated acute myeloid leukemia: 45-year follow up of a large family. Haematologica, 2016, 101, 846-852.	1.7	42
322	Genetic Complexity of Crohnâ€™s Disease in Two Large Ashkenazi Jewish Families. Gastroenterology, 2016, 151, 698-709.	0.6	54
323	A novel rasopathy caused by recurrent de novo missense mutations in PPP1CB closely resembles Noonan syndrome with loose anagen hair. American Journal of Medical Genetics, Part A, 2016, 170, 2237-2247.	0.7	117
324	A t(5;16) translocation is the likely driver of a syndrome with ambiguous genitalia, facial dysmorphism, intellectual disability, and speech delay. Journal of Physical Education and Sports Management, 2016, 2, a000703.	0.5	1
325	dbNSFP v3.0: A One-Stop Database of Functional Predictions and Annotations for Human Nonsynonymous and Splice-Site SNVs. Human Mutation, 2016, 37, 235-241.	1.1	845
326	Novel variants in ZNF34 and other brainâ€expressed transcription factors are shared among earlyâ€onset MDD relatives. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2016, 171, 333-341.	1.1	24
327	Pathogenic FBN1 variants in familial thoracic aortic aneurysms and dissections. Clinical Genetics, 2016, 89, 719-723.	1.0	26

#	Article	IF	Citations
328	<scp>WES</scp> in a family trio suggests involvement of <scp>TECPR2</scp> in a complex form of progressive motor neuron disease. Clinical Genetics, 2016, 90, 182-185.	1.0	17
329	dbWGFP: a database and web server of human whole-genome single nucleotide variants and their functional predictions. Database: the Journal of Biological Databases and Curation, 2016, 2016, baw024.	1.4	27
330	Whole-genome sequencing expands diagnostic utility and improves clinical management in paediatric medicine. Npj Genomic Medicine, 2016, 1, .	1.7	295
331	Whole genome sequence analysis of serum amino acid levels. Genome Biology, 2016, 17, 237.	3.8	17
332	New insights into the generation and role of de novo mutations in health and disease. Genome Biology, 2016, 17, 241.	3.8	339
333	Natural resistance to Meningococcal Disease related to CFH loci: Meta-analysis of genome-wide association studies. Scientific Reports, 2016, 6, 35842.	1.6	33
334	Functional assays provide a robust tool for the clinical annotation of genetic variants of uncertain significance. Npj Genomic Medicine, 2016, 1, .	1.7	70
335	In silico functional dissection of saturation mutagenesis: Interpreting the relationship between phenotypes and changes in protein stability, interactions and activity. Scientific Reports, 2016, 6, 19848.	1.6	87
336	Establishing the involvement of the novel geneAGBL5in retinitis pigmentosa by whole genome sequencing. Physiological Genomics, 2016, 48, 922-927.	1.0	29
337	Genetic, immunological, and clinical features of patients with bacterial and fungal infections due to inherited IL-17RA deficiency. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, E8277-E8285.	3.3	137
338	Understanding rare and common diseases in the context of human evolution. Genome Biology, 2016, 17, 225.	3.8	76
339	SCN8A mutation in a child presenting with seizures and developmental delays. Journal of Physical Education and Sports Management, 2016, 2, a001073.	0.5	12
340	iCAGES: integrated CAncer GEnome Score for comprehensively prioritizing driver genes in personal cancer genomes. Genome Medicine, 2016, 8, 135.	3.6	45
341	MIB2variants altering NOTCH signalling result in left ventricle hypertrabeculation/non-compaction and are associated with MÃ©nÃ©trier-like gastropathy. Human Molecular Genetics, 2016, 26, ddw365.	1.4	7
342	Genome sequencing in a case of Niemannâ€“Pick type C. Journal of Physical Education and Sports Management, 2016, 2, a001222.	0.5	10
343	A dominant gain-of-function mutation in universal tyrosine kinase SRC causes thrombocytopenia, myelofibrosis, bleeding, and bone pathologies. Science Translational Medicine, 2016, 8, 328ra30.	5.8	87
344	Increased burden of deleterious variants in essential genes in autism spectrum disorder. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 15054-15059.	3.3	68
345	A genome-wide analysis in cluster headache points to neprilysin and PACAP receptor gene variants. Journal of Headache and Pain, 2016, 17, 114.	2.5	38

#	Article	IF	Citations
346	Analysis of Plasminogen Genetic Variants in Multiple Sclerosis Patients. G3: Genes, Genomes, Genetics, 2016, 6, 2073-2079.	0.8	13
347	SVScore: an impact prediction tool for structural variation. Bioinformatics, 2017, 33, 1083-1085.	1.8	54
348	A childhood acute lymphoblastic leukemia genome-wide association study identifies novel sex-specific risk variants. Medicine (United States), 2016, 95, e5300.	0.4	20
349	Mutations inHECW2are associated with intellectual disability and epilepsy. Journal of Medical Genetics, 2016, 53, 697-704.	1.5	55
350	Applications of the 1000 Genomes Project resources. Briefings in Functional Genomics, 2017, 16, elw027.	1.3	30
351	Germ line mutations in shelterin complex genes are associated with familial chronic lymphocytic leukemia. Blood, 2016, 128, 2319-2326.	0.6	90
352	Uncovering disease mechanisms through network biology in the era of Next Generation Sequencing. Scientific Reports, 2016, 6, 24570.	1.6	29
353	Towards a molecular classification of pulmonary arterial hypertension. European Respiratory Journal, 2016, 48, 987-989.	3.1	2
354	iFish: predicting the pathogenicity of human nonsynonymous variants using gene-specific/family-specific attributes and classifiers. Scientific Reports, 2016, 6, 31321.	1.6	24
355	A novel ANO3 variant identified in a 53-year-old woman presenting with hyperkinetic dysarthria, blepharospasm, hyperkinesias, and complex motorÂtics. BMC Medical Genetics, 2016, 17, 93.	2.1	14
356	DeepEnhancer: Predicting enhancers by convolutional neural networks. , 2016, , .		9
357	The ONDRISeq panel: custom-designed next-generation sequencing of genes related to neurodegeneration. Npj Genomic Medicine, 2016, 1, 16032.	1.7	26
358	Codon bias and the folding dynamics of the cystic fibrosis transmembrane conductance regulator. Cellular and Molecular Biology Letters, 2016, 21, 23.	2.7	32
359	Non-Coding Loss-of-Function Variation in Human Genomes. Human Heredity, 2016, 81, 78-87.	0.4	16
360	UMDâ€Predictor: A Highâ€Throughput Sequencing Compliant System for Pathogenicity Prediction of any Human cDNA Substitution. Human Mutation, 2016, 37, 439-446.	1.1	104
361	IMPACT: a whole-exome sequencing analysis pipeline for integrating molecular profiles with actionable therapeutics in clinical samples. Journal of the American Medical Informatics Association: JAMIA, 2016, 23, 721-730.	2.2	38
362	On the phenotypic spectrum of serine biosynthesis defects. Journal of Inherited Metabolic Disease, 2016, 39, 373-381.	1.7	45
363	Exome sequencing reveals two novel compound heterozygous XYLT1 mutations in a Polish patient with Desbuquois dysplasia type 2 and growth hormone deficiency. Journal of Human Genetics, 2016, 61, 577-583.	1.1	18

#	Article	IF	Citations
364	When Is Selection Effective?. Genetics, 2016, 203, 451-462.	1.2	73
365	Identification of a mutation causing hypertrophic cardiomyopathy using whole exome sequencing: A proof-of-concept. Journal of Cardiology, 2016, 67, 131-132.	0.8	2
366	Decoding transcriptional enhancers: Evolving from annotation to functional interpretation. Seminars in Cell and Developmental Biology, 2016, 57, 40-50.	2.3	11
367	The genetic background of arrhythmogenic right ventricular cardiomyopathy. Journal of Arrhythmia, 2016, 32, 398-403.	0.5	58
368	Discovery of rare variants for complex phenotypes. Human Genetics, 2016, 135, 625-634.	1.8	40
369	Expanding the mutation spectrum in 130 probands with ARPKD: identification of 62 novel PKHD1 mutations by sanger sequencing and MLPA analysis. Journal of Human Genetics, 2016, 61, 811-821.	1.1	27
370	A nonsense mutation in the DNA repair factor Hebo causes mild bone marrow failure and microcephaly. Journal of Experimental Medicine, 2016, 213, 1011-1028.	4.2	34
371	A novel nonsense GPSM2 mutation in a Yemeni family underlying Chudley-McCullough syndrome. European Journal of Medical Genetics, 2016, 59, 337-341.	0.7	9
372	PANTHER-PSEP: predicting disease-causing genetic variants using position-specific evolutionary preservation. Bioinformatics, 2016, 32, 2230-2232.	1.8	228
373	MultisystemicSYNE1ataxia: confirming the high frequency and extending the mutational and phenotypic spectrum. Brain, 2016, 139, e46-e46.	3.7	40
374	Genetic cause and prevalence of hydroxyprolinemia. Journal of Inherited Metabolic Disease, 2016, 39, 625-632.	1.7	17
375	An extended set of yeast-based functional assays accurately identifies human disease mutations. Genome Research, 2016, 26, 670-680.	2.4	116
376	A Standardized DNA Variant Scoring System for Pathogenicity Assessments in Mendelian Disorders. Human Mutation, 2016, 37, 127-134.	1.1	52
377	Recessive Inactivating Mutations in TBCK, Encoding a Rab GTPase-Activating Protein, Cause Severe Infantile Syndromic Encephalopathy. American Journal of Human Genetics, 2016, 98, 772-781.	2.6	43
378	Enhancerâ€“promoter interactions are encoded by complex genomic signatures on looping chromatin. Nature Genetics, 2016, 48, 488-496.	9.4	369
379	De Novo Mutations in PDE10A Cause Childhood-Onset Chorea with Bilateral Striatal Lesions. American Journal of Human Genetics, 2016, 98, 763-771.	2.6	96
380	Hoyeraal-Hreidarsson Syndrome due to PARN Mutations: Fourteen Years of Follow-Up. Pediatric Neurology, 2016, 56, 62-68.e1.	1.0	29
381	Heterozygous colon cancer-associated mutations of SAMHD1 have functional significance. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 4723-4728.	3.3	100

#	Article	IF	Citations
382	Novel bioinformatic developments for exome sequencing. Human Genetics, 2016, 135, 603-614.	1.8	37
383	Ligase-4 Deficiency Causes Distinctive Immune Abnormalities in Asymptomatic Individuals. Journal of Clinical Immunology, 2016, 36, 341-353.	2.0	30
384	Biallelic BRCA2 Mutations Shape the Somatic Mutational Landscape of Aggressive Prostate Tumors. American Journal of Human Genetics, 2016, 98, 818-829.	2.6	34
385	A flexible method for estimating the fraction of fitness influencing mutations from large sequencing data sets. Genome Research, 2016, 26, 834-843.	2.4	10
386	SYNE1 ataxia is a common recessive ataxia with major non-cerebellar features: a large multi-centre study. Brain, 2016, 139, 1378-1393.	3.7	87
387	Genotype and phenotype in patients with Noonan syndrome and a RIT1 mutation. Genetics in Medicine, 2016, 18, 1226-1234.	1.1	77
388	CADD score has limited clinical validity for the identification of pathogenic variants in noncoding regions in a hereditary cancer panel. Genetics in Medicine, 2016, 18, 1269-1275.	1.1	45
389	Familial early-onset dementia with complex neuropathologic phenotype and genomic background. Neurobiology of Aging, 2016, 42, 199-204.	1.5	16
390	Comparison of pathogenicity prediction tools on missense variants in RYR1 and CACNA1S associated with malignant hyperthermia. British Journal of Anaesthesia, 2016, 117, 124-128.	1.5	34
391	Association Tests for Rare Variants. Annual Review of Genomics and Human Genetics, 2016, 17, 117-130.	2.5	56
392	Identification of Common Genetic Variants Influencing Spontaneous Dizygotic Twinning and Female Fertility. American Journal of Human Genetics, 2016, 98, 898-908.	2.6	89
393	CSF1Rmosaicism in a family with hereditary diffuse leukoencephalopathy with spheroids. Brain, 2016, 139, 1666-1672.	3.7	53
394	Carrier Screening is a Deficient Strategy for Determining Sperm Donor Eligibility and Reducing Risk of Disease in Recipient Children. Genetic Testing and Molecular Biomarkers, 2016, 20, 276-284.	0.3	23
395	Punctuated bursts in human male demography inferred from 1,244 worldwide Y-chromosome sequences. Nature Genetics, 2016, 48, 593-599.	9.4	273
396	A Broad Overview of Computational Methods for Predicting the Pathophysiological Effects of Non-synonymous Variants. Methods in Molecular Biology, 2016, 1415, 423-440.	0.4	7
397	Hematopoietic stem cell transplantation for CTLA4 deficiency. Journal of Allergy and Clinical Immunology, 2016, 138, 615-619.e1.	1.5	88
398	Separating the wheat from the chaff: systematic identification of functionally relevant noncoding variants in ADHD. Molecular Psychiatry, 2016, 21, 1589-1598.	4.1	7
399	Germline De Novo Mutations in GNB1 Cause Severe Neurodevelopmental Disability, Hypotonia, and Seizures. American Journal of Human Genetics, 2016, 98, 1001-1010.	2.6	102

#	Article	IF	Citations
400	Comment: A Human Genetics Perspective. Journal of the American Statistical Association, 2016, 111, 124-127.	1.8	1
401	How to Identify Pathogenic Mutations among All Those Variations: Variant Annotation and Filtration in the Genome Sequencing Era. Human Mutation, 2016, 37, 1272-1282.	1.1	28
402	De novo PHIP-predicted deleterious variants are associated with developmental delay, intellectual disability, obesity, and dysmorphic features. Journal of Physical Education and Sports Management, 2016, 2, a001172.	0.5	42
403	Familial occurrence of brain arteriovenous malformation: a novel ACVRL1 mutation detected by whole exome sequencing. Journal of Neurosurgery, 2016, 126, 1879-1883.	0.9	16
404	Bioinformatics and Orphan Diseases. Translational Bioinformatics, 2016, , 313-338.	0.0	0
406	Recurrent De Novo Dominant Mutations in SLC25A4 Cause Severe Early-Onset Mitochondrial Disease and Loss of Mitochondrial DNA Copy Number. American Journal of Human Genetics, 2016, 99, 860-876.	2.6	93
407	A unified analytic framework for prioritization of non-coding variants of uncertain significance in heritable breast and ovarian cancer. BMC Medical Genomics, 2016, 9, 19.	0.7	28
408	Deep sequencing of 10,000 human genomes. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 11901-11906.	3.3	315
409	Exome and genome sequencing for inborn errors of immunity. Journal of Allergy and Clinical Immunology, 2016, 138, 957-969.	1.5	187
410	Atopic Dermatitis Susceptibility Variants in FilaggrinHitchhikeHornerin Selective Sweep. Genome Biology and Evolution, 2016, 8, 3240-3255.	1.1	35
411	Mutational landscape of prostate tumors revealed by whole-exome sequencing. Russian Journal of Genetics, 2016, 52, 999-1003.	0.2	1
412	The genetics of breast cancer risk in the post-genome era: thoughts on study design to move past BRCA and towards clinical relevance. Breast Cancer Research, 2016, 18, 99.	2.2	72
413	Chromatin structureâ€“based prediction of recurrent noncoding mutations in cancer. Nature Genetics, 2016, 48, 1321-1326.	9.4	29
414	The role of next generation sequencing in understanding male and female sexual development: clinical implications. Expert Review of Endocrinology and Metabolism, 2016, 11, 433-443.	1.2	3
416	The Ensembl Variant Effect Predictor. Genome Biology, 2016, 17, 122.	3.8	5,181
417	REVEL: An Ensemble Method for Predicting the Pathogenicity of Rare Missense Variants. American Journal of Human Genetics, 2016, 99, 877-885.	2.6	1,555
418	Genetic Factors of the Disease Course after Sepsis: A Genome-Wide Study for 28 Day Mortality. EBioMedicine, 2016, 12, 239-246.	2.7	52
419	Biallelic Mutations in TBCD , Encoding the Tubulin Folding Cofactor D, Perturb Microtubule Dynamics and Cause Early-Onset Encephalopathy. American Journal of Human Genetics, 2016, 99, 962-973.	2.6	66

#	Article	IF	Citations
420	TBCE Mutations Cause Early-Onset Progressive Encephalopathy with Distal Spinal Muscular Atrophy. American Journal of Human Genetics, 2016, 99, 974-983.	2.6	49
421	Guidelines for Large-Scale Sequence-Based Complex Trait Association Studies: Lessons Learned from the NHLBI Exome Sequencing Project. American Journal of Human Genetics, 2016, 99, 791-801.	2.6	79
422	Weighted K-means support vector machine for cancer prediction. SpringerPlus, 2016, 5, 1162.	1.2	23
423	A Pleiotropic Missense Variant in SLC39A8 Is Associated With Crohnâ€™s Disease and Human Gut Microbiome Composition. Gastroenterology, 2016, 151, 724-732.	0.6	109
424	An inactivating mutation in intestinal cell kinase,ICK, impairs hedgehog signalling and causes short rib-polydactyly syndrome. Human Molecular Genetics, 2016, 25, 3998-4011.	1.4	44
425	Phenotypic insights into ADCY5 â€associated disease. Movement Disorders, 2016, 31, 1033-1040.	2.2	106
426	Beta thalassemia in 31,734 cases with HBB gene mutations: Pathogenic and structural analysis of the common mutations; Iran as the crossroads of the Middle East. Blood Reviews, 2016, 30, 493-508.	2.8	31
427	Genetic variation among 82 pharmacogenes: The PGRNseq data from the eMERGE network. Clinical Pharmacology and Therapeutics, 2016, 100, 160-169.	2.3	163
428	Hutteriteâ€type cataract maps to chromosome 6p21.32â€p21.31, cosegregates with a homozygous mutation in <scp>LEMD</scp>2, and is associated with sudden cardiac death. Molecular Genetics & Genomic Medicine, 2016, 4, 77-94.	0.6	28
429	Computational assessment of feature combinations for pathogenic variant prediction. Molecular Genetics & Genomic Medicine, 2016, 4, 431-446.	0.6	13
430	Importance of Genetic Studies in Consanguineous Populations for the Characterization of Novel Human Gene Functions. Annals of Human Genetics, 2016, 80, 187-196.	0.3	41
431	Tentative clinical diagnosis of Lujanâ€Fryns syndromeâ€”A conglomeration of different genetic entities?. American Journal of Medical Genetics, Part A, 2016, 170, 94-102.	0.7	11
432	Acadian variant of Fanconi syndrome is caused by mitochondrial respiratory chain complex I deficiency due to a non-coding mutation in complex I assembly factor NDUFAF6. Human Molecular Genetics, 2016, 25, 4062-4079.	1.4	55
433	A Whole-Genome Analysis Framework for Effective Identification of Pathogenic Regulatory Variants in Mendelian Disease. American Journal of Human Genetics, 2016, 99, 595-606.	2.6	223
434	Implementation of next-generation sequencing for molecular diagnosis of hereditary breast and ovarian cancer highlights its genetic heterogeneity. Breast Cancer Research and Treatment, 2016, 159, 245-256.	1.1	23
435	Acute Intermittent Porphyria: Predicted Pathogenicity ofHMBSVariants Indicates Extremely Low Penetrance of the Autosomal Dominant Disease. Human Mutation, 2016, 37, 1215-1222.	1.1	129
436	Spectrum of Mutations in Hypertrophic Cardiomyopathy Genes Among Tunisian Patients. Genetic Testing and Molecular Biomarkers, 2016, 20, 674-679.	0.3	11
437	Loss of function of SLC25A46 causes lethal congenital pontocerebellar hypoplasia. Brain, 2016, 139, 2877-2890.	3.7	74

#	Article	IF	Citations
438	Reevaluation of RINT1 as a breast cancer predisposition gene. Breast Cancer Research and Treatment, 2016, 159, 385-392.	1.1	16
439	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
440	Mutational Spectrum in Holoprosencephaly Shows That FGF is a New Major Signaling Pathway. Human Mutation, 2016, 37, 1329-1339.	1.1	56
441	Inheritance-mode specific pathogenicity prioritization (ISPP) for human protein coding genes. Bioinformatics, 2016, 32, 3065-3071.	1.8	11
442	The Heritable Basis of Congenital Heart Disease. Circulation: Cardiovascular Genetics, 2016, 9, 315-317.	5.1	4
443	Targeted sequencing of 351 candidate genes for epileptic encephalopathy in a large cohort of patients. Molecular Genetics & Genomic Medicine, 2016, 4, 568-580.	0.6	83
444	IMHOTEPâ€"a composite score integrating popular tools for predicting the functional consequences of non-synonymous sequence variants. Nucleic Acids Research, 2017, 45, gkw886.	6.5	10
445	Polygenic Versus Monogenic Causes of Hypercholesterolemia Ascertained Clinically. Arteriosclerosis, Thrombosis, and Vascular Biology, 2016, 36, 2439-2445.	1.1	174
446	The Qatar genome: a population-specific tool for precision medicine in the Middle East. Human Genome Variation, 2016, 3, 16016.	0.4	103
448	Combined variants in factor VIII and prostaglandin synthase-1 amplify hemorrhage severity across three generations of descendants. Journal of Thrombosis and Haemostasis, 2016, 14, 2230-2240.	1.9	6
449	Actionable Genes, Core Databases, and Locus-Specific Databases. Human Mutation, 2016, 37, 1299-1307.	1.1	6
450	A de novo missense mutation in ZMYND11 is associated with global developmental delay, seizures, and hypotonia. Journal of Physical Education and Sports Management, 2016, 2, a000851.	0.5	16
451	WES/WGS Reporting of Mutations from Cardiovascular â€œActionableâ€•Genes in Clinical Practice: A Key Role for UMD Knowledgebases in the Era of Big Databases. Human Mutation, 2016, 37, 1308-1317.	1.1	5
452	Mutations in Human Accelerated Regions Disrupt Cognition and Social Behavior. Cell, 2016, 167, 341-354.e12.	13.5	280
453	Dual T cellâ€“ and B cellâ€“intrinsic deficiency in humans with biallelic RLTPR mutations. Journal of Experimental Medicine, 2016, 213, 2413-2435.	4.2	117
454	Translational plasticity facilitates the accumulation of nonsense genetic variants in the human population. Genome Research, 2016, 26, 1639-1650.	2.4	31
455	Unsupervised detection of cancer driver mutations with parsimony-guided learning. Nature Genetics, 2016, 48, 1288-1294.	9.4	52
456	De Novo Mutations in CHD4 , an ATP-Dependent Chromatin Remodeler Gene, Cause an Intellectual Disability Syndrome with Distinctive Dysmorphisms. American Journal of Human Genetics, 2016, 99, 934-941.	2.6	111

#	Article	IF	Citations
457	Mutations in Citron Kinase Cause Recessive Microlissencephaly with Multinucleated Neurons. American Journal of Human Genetics, 2016, 99, 511-520.	2.6	59
458	De novo nonsense and frameshift variants of TCF20 in individuals with intellectual disability and postnatal overgrowth. European Journal of Human Genetics, 2016, 24, 1739-1745.	1.4	28
459	Association of kidney structure-related gene variants with type 2 diabetes-attributed end-stage kidney disease in African Americans. Human Genetics, 2016, 135, 1251-1262.	1.8	43
460	Regulatory Single-Nucleotide Variant Predictor Increases Predictive Performance of Functional Regulatory Variants. Human Mutation, 2016, 37, 1137-1143.	1.1	13
461	Bioinformatics in Neuropsychiatric Genomics. , 2016, , 93-100.		0
462	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042.	9.4	218
463	Truncating and missense PPM1D mutations in earlyâ€onset and/or familial/hereditary prostate cancer patients. Genes Chromosomes and Cancer, 2016, 55, 954-961.	1.5	15
464	The Complementarity Between Protein-Specific and General Pathogenicity Predictors for Amino Acid Substitutions. Human Mutation, 2016, 37, 1013-1024.	1.1	42
465	Genetic and clinical characterization of Pakistani families with Bardet-Biedl syndrome extends the genetic and phenotypic spectrum. Scientific Reports, 2016, 6, 34764.	1.6	29
466	The population genomics of rhesus macaques (Macaca mulatta) based on whole-genome sequences. Genome Research, 2016, 26, 1651-1662.	2.4	101
467	Mutational profile of primary dermal melanoma: A case series. Journal of the American Academy of Dermatology, 2016, 75, 1263-1265.e5.	0.6	7
468	Genome-wide association study identifies multiple susceptibility loci for multiple myeloma. Nature Communications, 2016, 7, 12050.	5.8	146
469	Pedigree based DNA sequencing pipeline for germline genomes of cancer families. Hereditary Cancer in Clinical Practice, 2016, 14, 16.	0.6	7
470	Current Perspectives in Autism Spectrum Disorder: From Genes to Therapy. Journal of Neuroscience, 2016, 36, 11402-11410.	1.7	44
471	The use of targeted exome sequencing in genetic diagnosis of young patients with severe hypercholesterolemia. Scientific Reports, 2016, 6, 36823.	1.6	13
472	Prioritization of non-coding disease-causing variants and long non-coding RNAs in liver cancer. Oncology Letters, 2016, 12, 3987-3994.	0.8	8
473	A continuum of admixture in the Western Hemisphere revealed by the African Diaspora genome. Nature Communications, 2016, 7, 12522.	5.8	136
474	Genome-wide significance testing of variation from single case exomes. Nature Genetics, 2016, 48, 1455-1461.	9.4	43

#	Article	IF	Citations
475	M-CAP eliminates a majority of variants of uncertain significance in clinical exomes at high sensitivity. Nature Genetics, 2016, 48, 1581-1586.	9.4	654
476	RASGRP1 deficiency causes immunodeficiency with impaired cytoskeletal dynamics. Nature Immunology, 2016, 17, 1352-1360.	7.0	115
477	Landscape of warfarin and clopidogrel pharmacogenetic variants in Qatari population from whole exome datasets. Pharmacogenomics, 2016, 17, 1891-1901.	0.6	14
478	Big Data Analytics in Genomics. , 2016, , .		7
479	Challenges and disparities in the application of personalized genomic medicine to populations with African ancestry. Nature Communications, 2016, 7, 12521.	5.8	68
480	The structural origin of metabolic quantitative diversity. Scientific Reports, 2016, 6, 31463.	1.6	18
481	Seemingly dominant inheritance of a recessiveANO10mutation in romani families with cerebellar ataxia. Movement Disorders, 2016, 31, 1929-1931.	2.2	12
482	Genomic Variants Associated with Resistance to High Fat Diet Induced Obesity in a Primate Model. Scientific Reports, 2016, 6, 36123.	1.6	23
483	SSA-ME Detection of cancer driver genes using mutual exclusivity by small subnetwork analysis. Scientific Reports, 2016, 6, 36257.	1.6	12
484	Germline and somatic mutations in the MTOR gene in focal cortical dysplasia and epilepsy. Neurology: Genetics, 2016, 2, e118.	0.9	125
485	Genome-wide characteristics of de novo mutations in autism. Npj Genomic Medicine, 2016, 1, 160271-1602710.	1.7	200
486	Classification of breast cancer patients using somatic mutation profiles and machine learning approaches. BMC Systems Biology, 2016, 10, 62.	3.0	54
487	NGS for Sequence Variants. Advances in Experimental Medicine and Biology, 2016, 939, 1-20.	0.8	2
488	Loss of RNA expression and allele-specific expression associated with congenital heart disease. Nature Communications, 2016, 7, 12824.	5.8	51
489	OncoMiner: A Pipeline for Bioinformatics Analysis of Exonic Sequence Variants in Cancer. , 2016, , 373-396.		2
490	Deep-targeted exon sequencing reveals renal polymorphisms associate with postexercise hypotension among African Americans. Physiological Reports, 2016, 4, e12992.	0.7	8
491	KBG syndrome involving a single-nucleotide duplication in ANKRD11. Journal of Physical Education and Sports Management, 2016, 2, a001131.	0.5	15
492	Global inference of disease-causing single nucleotide variants from exome sequencing data. BMC Bioinformatics, 2016, 17, 468.	1.2	4

#	Article	IF	Citations
493	Targeted high-throughput sequencing of candidate genes for chronic obstructive pulmonary disease. BMC Pulmonary Medicine, 2016, 16, 146.	0.8	12
494	Incorporating ENCODE information into association analysis of whole genome sequencing data. BMC Proceedings, 2016, 10, 257-261.	1.8	9
495	Identity-by-descent mapping for diastolic blood pressure in unrelated Mexican Americans. BMC Proceedings, 2016, 10, 263-267.	1.8	4
496	DIVAN: accurate identification of non-coding disease-specific risk variants using multi-omics profiles. Genome Biology, 2016, 17, 252.	3.8	67
497	Clinical Implementation of Integrated Genomic Profiling in Patients with Advanced Cancers. Scientific Reports, 2016, 6, 25.	1.6	32
498	KinMutRF: a random forest classifier of sequence variants in the human protein kinase superfamily. BMC Genomics, 2016, 17, 396.	1.2	11
499	Explorations to improve the completeness of exome sequencing. BMC Medical Genomics, 2016, 9, 56.	0.7	9
500	Immunoseq: the identification of functionally relevant variants through targeted capture and sequencing of active regulatory regions in human immune cells. BMC Medical Genomics, 2016, 9, 59.	0.7	26
501	Retinoic acid catabolizing enzyme CYP 26C1 is a genetic modifier in SHOX deficiency. EMBO Molecular Medicine, 2016, 8, 1455-1469.	3.3	23
502	The discovery of LRRK2 p.R1441S, a novel mutation for Parkinson's disease, adds to the complexity of a mutational hotspot. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2016, 171, 925-930.	1.1	17
503	Whole exome sequencing is necessary to clarify ID/DD cases with de novo copy number variants of uncertain significance: Two proofâ€ofâ€concept examples. American Journal of Medical Genetics, Part A, 2016, 170, 1772-1779.	0.7	26
504	Characterization of patients at high risk of melanoma in Austria. British Journal of Dermatology, 2016, 174, 1308-1317.	1.4	10
505	Tools for Predicting the Functional Impact of Nonsynonymous Genetic Variation. Genetics, 2016, 203, 635-647.	1.2	84
506	Family-based approaches: design, imputation, analysis, and beyond. BMC Genetics, 2016, 17, 9.	2.7	13
507	Predicting regulatory variants with composite statistic. Bioinformatics, 2016, 32, 2729-2736.	1.8	40
509	ABCA7 frameshift deletion associated with Alzheimer disease in African Americans. Neurology: Genetics, 2016, 2, e79.	0.9	74
510	The genetic heterogeneity of colorectal cancer predisposition - guidelines for gene discovery. Cellular Oncology (Dordrecht), 2016, 39, 491-510.	2.1	34
512	The structural effects of mutations can aid in differential phenotype prediction of beta-myosin heavy chain (Myosin-7) missense variants. Bioinformatics, 2016, 32, 2947-2955.	1.8	9

#	Article	IF	Citations
513	Basset: learning the regulatory code of the accessible genome with deep convolutional neural networks. Genome Research, 2016, 26, 990-999.	2.4	790
514	Platelet-Related Variants Identified by Exomechip Meta-analysis in 157,293 Individuals. American Journal of Human Genetics, 2016, 99, 40-55.	2.6	82
515	Mitochondrial Complex III Deficiency with Ketoacidosis and Hyperglycemia Mimicking Neonatal Diabetes. JIMD Reports, 2016, 31, 57-62.	0.7	5
516	Prospecting for Precision: Promises for Personalized Medicine. Journal of Rheumatology, 2016, 43, 999-1000.	1.0	2
517	Rare variants in dementia genes and Parkinsonâ€™s disease. European Journal of Human Genetics, 2016, 24, 1661-1662.	1.4	0
518	Whole genome sequencing and its applications in medical genetics. Quantitative Biology, 2016, 4, 115-128.	0.3	7
519	Filtering genetic variants and placing informative priors based on putative biological function. BMC Genetics, 2016, 17, 8.	2.7	6
520	Pathogenic copy number variants and SCN1A mutations in patients with intellectual disability and childhood-onset epilepsy. BMC Medical Genetics, 2016, 17, 34.	2.1	23
521	Mitochondrial DNA variants correlate with symptoms in myalgic encephalomyelitis/chronic fatigue syndrome. Journal of Translational Medicine, 2016, 14, 19.	1.8	42
522	High-performance web services for querying gene and variant annotation. Genome Biology, 2016, 17, 91.	3.8	166
523	Neuronal deficiency ofARV1causes an autosomal recessive epileptic encephalopathy. Human Molecular Genetics, 2016, 25, ddw157.	1.4	23
524	OncodriveFML: a general framework to identify coding and non-coding regions with cancer driver mutations. Genome Biology, 2016, 17, 128.	3.8	251
525	Mitral regurgitation as a phenotypic manifestation of nonphotosensitive trichothiodystrophy due to a splice variant in MPLKIP. BMC Medical Genetics, 2016, 17, 13.	2.1	5
526	Phenotype and genotype in patients with Larsen syndrome: clinical homogeneity and allelic heterogeneity in seven patients. BMC Medical Genetics, 2016, 17, 27.	2.1	18
527	Interactive Exploration, Analysis, and Visualization of Complex Phenome-Genome Datasets with ASPIREdb. Human Mutation, 2016, 37, 719-726.	1.1	2
528	In Silico Functional Annotation of Genomic Variation. Current Protocols in Human Genetics, 2016, 88, 6.15.1-6.15.17.	3.5	20
529	Associations Between Common and Rare Exonic Genetic Variants and Serum Levels of 20 Cardiovascular-Related Proteins. Circulation: Cardiovascular Genetics, 2016, 9, 375-383.	5.1	18
530	Large-Scale Exome-wide Association Analysis Identifies Loci for White Blood Cell Traits and Pleiotropy with Immune-Mediated Diseases. American Journal of Human Genetics, 2016, 99, 22-39.	2.6	50

#	Article	IF	Citations
531	X-exome sequencing of 405 unresolved families identifies seven novel intellectual disability genes. Molecular Psychiatry, 2016, 21, 133-148.	4.1	243
532	NovelSCN10Avariants associated with Brugada syndrome. Europace, 2016, 18, 905-911.	0.7	31
533	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
534	Mutational analysis of TBK1 in Taiwanese patients with amyotrophic lateral sclerosis. Neurobiology of Aging, 2016, 40, 191.e11-191.e16.	1.5	46
535	Overlap between Parkinson disease and Alzheimer disease in ABCA7 functional variants. Neurology: Genetics, 2016, 2, e44.	0.9	31
536	The mutation significance cutoff: gene-level thresholds for variant predictions. Nature Methods, 2016, 13, 109-110.	9.0	249
537	The intolerance to functional genetic variation of protein domains predicts the localization of pathogenic mutations within genes. Genome Biology, 2016, 17, 9.	3.8	118
538	A novel human autoimmune syndrome caused by combined hypomorphic and activating mutations in ZAP-70. Journal of Experimental Medicine, 2016, 213, 155-165.	4.2	83
539	A Multiplex Kindred with Hennekam Syndrome due to Homozygosity for a CCBE1 Mutation that does not Prevent Protein Expression. Journal of Clinical Immunology, 2016, 36, 19-27.	2.0	12
540	Exome Array Analysis Identifies a Common Variant in IL27 Associated with Chronic Obstructive Pulmonary Disease. American Journal of Respiratory and Critical Care Medicine, 2016, 194, 48-57.	2.5	52
541	Role of non-coding sequence variants in cancer. Nature Reviews Genetics, 2016, 17, 93-108.	7.7	420
542	GenoWAP: GWAS signal prioritization through integrated analysis of genomic functional annotation. Bioinformatics, 2016, 32, 542-548.	1.8	47
543	Non-coding genome functions in diabetes. Journal of Molecular Endocrinology, 2016, 56, R1-R20.	1.1	12
544	Desmoplakin Variants Are Associated with Idiopathic Pulmonary Fibrosis. American Journal of Respiratory and Critical Care Medicine, 2016, 193, 1151-1160.	2.5	68
545	Secondary neurotransmitter deficiencies in epilepsy caused by voltage-gated sodium channelopathies: A potential treatment target?. Molecular Genetics and Metabolism, 2016, 117, 42-48.	0.5	40
546	Exome sequencing explained: a practical guide to its clinical application. Briefings in Functional Genomics, 2016, 15, 374-384.	1.3	58
547	Testing for Ancient Selection Using Cross-population Allele Frequency Differentiation. Genetics, 2016, 202, 733-750.	1.2	101
548	Robust classification of protein variation using structural modelling and large-scale data integration. Nucleic Acids Research, 2016, 44, 2501-2513.	6.5	52

#	Article	IF	Citations
549	Cytosolic phosphoenolpyruvate carboxykinase deficiency presenting with acute liver failure following gastroenteritis. Molecular Genetics and Metabolism, 2016, 118, 21-27.	0.5	23
550	Human Knockout Carriers: Dead, Diseased, Healthy, or Improved?. Trends in Molecular Medicine, 2016, 22, 341-351.	3.5	31
551	Novel mutations in LRP6 highlight the role of WNT signaling in tooth agenesis. Genetics in Medicine, 2016, 18, 1158-1162.	1.1	58
552	Developing maps of fitness consequences for plant genomes. Current Opinion in Plant Biology, 2016, 30, 101-107.	3.5	13
553	Multilevel biological characterization of exomic variants at the protein level significantly improves the identification of their deleterious effects. Bioinformatics, 2016, 32, 1797-1804.	1.8	32
554	Perspectives in Polycystic Ovary Syndrome: From Hair to Eternity. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 759-768.	1.8	71
555	A GABBR2 gene variant modifies pathophysiology in Huntingtonâ€™s disease. Neuroscience Letters, 2016, 620, 8-13.	1.0	13
556	Assessing the Pathogenicity of Insertion and Deletion Variants with the Variant Effect Scoring Tool (VESTâ€indel). Human Mutation, 2016, 37, 28-35.	1.1	101
557	Exome and genome sequencing: a revolution for the discovery and diagnosis of monogenic disorders. Journal of Internal Medicine, 2016, 279, 3-15.	2.7	71
558	Integrating population variation and protein structural analysis to improve clinical interpretation of missense variation: application to the WD40 domain. Human Molecular Genetics, 2016, 25, 927-935.	1.4	26
559	Focused Analysis of Exome Sequencing Data for Rare Germline Mutations in Familial and Sporadic Lung Cancer. Journal of Thoracic Oncology, 2016, 11, 52-61.	0.5	27
560	Commentary: Post-GWAS era: What can we do beyond cancer genetic association studies?. International Journal of Epidemiology, 2016, 45, 221-222.	0.9	1
561	The role of mutations in COL6A3 in isolated dystonia. Journal of Neurology, 2016, 263, 730-734.	1.8	15
562	Whole-genome mutational burden analysis of three pluripotency induction methods. Nature Communications, 2016, 7, 10536.	5.8	109
563	The effect of non-coding DNA variations on P53 and cMYC competitive inhibition at cis-overlapping motifs. Human Molecular Genetics, 2016, 25, 1517-1527.	1.4	7
564	Weighting sequence variants based on their annotation increases power of whole-genome association studies. Nature Genetics, 2016, 48, 314-317.	9.4	178
565	Multigene testing of moderate-risk genes: be mindful of the missense. Journal of Medical Genetics, 2016, 53, 366-376.	1.5	32
566	Effectiveness of whole-exome sequencing and costs of the traditional diagnostic trajectory in children with intellectual disability. Genetics in Medicine, 2016, 18, 949-956.	1.1	148

#	Article	IF	Citations
567	Mutations inEXOSC2are associated with a novel syndrome characterised by retinitis pigmentosa, progressive hearing loss, premature ageing, short stature, mild intellectual disability and distinctive gestalt. Journal of Medical Genetics, 2016, 53, 419-425.	1.5	69
568	Novel Insights into 46,XY Disorders of Sex Development due to <i>NR5A1</i> Gene Mutation. Sexual Development, 2015, 9, 260-268.	1.1	17
569	One SNP at a Time: Moving beyond GWAS in Psoriasis. Journal of Investigative Dermatology, 2016, 136, 567-573.	0.3	48
570	Haplotyping germline and cancer genomes with high-throughput linked-read sequencing. Nature Biotechnology, 2016, 34, 303-311.	9.4	617
571	Recent Selection Changes in Human Genes under Long-Term Balancing Selection. Molecular Biology and Evolution, 2016, 33, 1435-1447.	3.5	33
572	Genome-wide Association Study of Platelet Count Identifies Ancestry-Specific Loci in Hispanic/Latino Americans. American Journal of Human Genetics, 2016, 98, 229-242.	2.6	71
573	Identification of Gender-Specific Genetic Variants in Patients With Bicuspid Aortic Valve. American Journal of Cardiology, 2016, 117, 420-426.	0.7	53
574	Rare loss-of-function variants in SETD1A are associated with schizophrenia and developmental disorders. Nature Neuroscience, 2016, 19, 571-577.	7.1	388
575	No evidence that protein truncating variants inBRIP1are associated with breast cancer risk: implications for gene panel testing. Journal of Medical Genetics, 2016, 53, 298-309.	1.5	94
576	Risky business: getting a grip on BRIP. Journal of Medical Genetics, 2016, 53, 296-297.	1.5	1
577	Visual Prognosis in USH2A-Associated Retinitis Pigmentosa Is Worse for Patients with Usher Syndrome Type IIa Than for Those with Nonsyndromic Retinitis Pigmentosa. Ophthalmology, 2016, 123, 1151-1160.	2.5	76
578	A Statistical Approach for Testing Cross-Phenotype Effects of Rare Variants. American Journal of Human Genetics, 2016, 98, 525-540.	2.6	75
579	A novelHSD17B10mutation impairing the activities of the mitochondrial RNase P complex causes X-linked intractable epilepsy and neurodevelopmental regression. RNA Biology, 2016, 13, 477-485.	1.5	42
580	Complex inheritance of ABCA4 disease: four mutations in a family with multiple macular phenotypes. Human Genetics, 2016, 135, 9-19.	1.8	39
581	The promise and peril of genomic screening in the general population. Genetics in Medicine, 2016, 18, 593-599.	1.1	53
582	Integrating 400 million variants from 80,000 human samples with extensive annotations: towards a knowledge base to analyze disease cohorts. BMC Bioinformatics, 2016, 17, 24.	1.2	13
583	A spectral approach integrating functional genomic annotations for coding and noncoding variants. Nature Genetics, 2016, 48, 214-220.	9.4	506
584	The role of next-generation sequencing in understanding the genomic basis of diffuse large B cell lymphoma and advancing targeted therapies. Expert Review of Hematology, 2016, 9, 255-269.	1.0	12

#	Article	IF	Citations
585	GWASdb v2: an update database for human genetic variants identified by genome-wide association studies. Nucleic Acids Research, 2016, 44, D869-D876.	6.5	184
586	Autoimmune diseases â€" connecting risk alleles with molecular traits of the immune system. Nature Reviews Genetics, 2016, 17, 160-174.	7.7	173
587	Machine Learning in Genomic Medicine: A Review of Computational Problems and Data Sets. Proceedings of the IEEE, 2016, 104, 176-197.	16.4	186
588	Exome Sequencing Analysis in Severe, Early-Onset Chronic Obstructive Pulmonary Disease. American Journal of Respiratory and Critical Care Medicine, 2016, 193, 1353-1363.	2.5	46
589	Genomic Signatures of Selective Pressures and Introgression from Archaic Hominins at Human Innate Immunity Genes. American Journal of Human Genetics, 2016, 98, 5-21.	2.6	243
590	Pilot whole-exome sequencing of a German early-onset Alzheimer's disease cohort reveals a substantial frequency of PSEN2 variants. Neurobiology of Aging, 2016, 37, 208.e11-208.e17.	1.5	38
591	Pan-cancer analysis of the extent and consequences of intratumor heterogeneity. Nature Medicine, 2016, 22, 105-113.	15.2	629
592	LMNA cardiomyopathy detected in Japanese arrhythmogenic right ventricular cardiomyopathy cohort. Journal of Cardiology, 2016, 68, 346-351.	0.8	22
593	The Variant p.(Arg183Trp) in SPTLC2 Causes Late-Onset Hereditary Sensory Neuropathy. NeuroMolecular Medicine, 2016, 18, 81-90.	1.8	18
594	Whole-Exome Sequencing in Familial Parkinson Disease. JAMA Neurology, 2016, 73, 68.	4.5	71
595	Candidate DNA repair susceptibility genes identified by exome sequencing in high-risk pancreatic cancer. Cancer Letters, 2016, 370, 302-312.	3.2	47
596	Using Whole-Exome Sequencing to Identify Genetic Markers for Carboplatin and Gemcitabine-Induced Toxicities. Clinical Cancer Research, 2016, 22, 366-373.	3.2	20
597	Whole exome sequencing combined with integrated variant annotation prediction identifies a causative myosin essential light chain variant in hypertrophic cardiomyopathy. Journal of Cardiology, 2016, 67, 133-139.	0.8	14
598	De novo dominant variants affecting the motor domain of KIF1A are a cause of PEHO syndrome. European Journal of Human Genetics, 2016, 24, 949-953.	1.4	37
599	Targeted massively parallel sequencing of a panel of putative breast cancer susceptibility genes in a large cohort of multiple-case breast and ovarian cancer families. Journal of Medical Genetics, 2016, 53, 34-42.	1.5	63
600	Prioritization and burden analysis of rare variants in 208 candidate genes suggest they do not play a major role in CAKUT. Kidney International, 2016, 89, 476-486.	2.6	78
601	Interpreting functional effects of coding variants: challenges in proteome-scale prediction, annotation and assessment. Briefings in Bioinformatics, 2016, 17, 841-862.	3.2	23
602	Europeans have a higher proportion of high-frequency deleterious variants than Africans. Human Genetics, 2016, 135, 1-7.	1.8	15

#	Article	IF	Citations
603	A Molecular Evolutionary Reference for the Human Variome. Molecular Biology and Evolution, 2016, 33, 245-254.	3.5	20
604	Leveraging ancestry to improve causal variant identification in exome sequencing for monogenic disorders. European Journal of Human Genetics, 2016, 24, 113-119.	1.4	3
605	A novel missense mutation in ATRX uncovered in a Yemeni family leads to alpha-thalassemia/mental retardation syndrome without alpha-thalassemia. Irish Journal of Medical Science, 2017, 186, 333-337.	0.8	5
606	Mutations in the <scp>IRBIT</scp> domain of <scp>ITPR1</scp> are a frequent cause of autosomal dominant nonprogressive congenital ataxia. Clinical Genetics, 2017, 91, 86-91.	1.0	30
607	In SilicoFunctional Meta-Analysis of 5,962ABCA4Variants in 3,928 Retinal Dystrophy Cases. Human Mutation, 2017, 38, 400-408.	1.1	118
608	iRegNet3D: three-dimensional integrated regulatory network for the genomic analysis of coding and non-coding disease mutations. Genome Biology, 2017, 18, 10.	3.8	9
609	Functional and clinical relevance of novel and known PCSK1 variants for childhood obesity and glucose metabolism. Molecular Metabolism, 2017, 6, 295-305.	3.0	26
610	Investigating the role of ALS genes CHCHD10 and TUBA4A in Belgian FTD-ALS spectrum patients. Neurobiology of Aging, 2017, 51, 177.e9-177.e16.	1.5	60
611	Combined alpha-delta platelet storage pool deficiency is associated with mutations in GFI1B. Molecular Genetics and Metabolism, 2017, 120, 288-294.	0.5	22
612	EIF2S3Mutations Associated with Severe X-Linked Intellectual Disability Syndrome MEHMO. Human Mutation, 2017, 38, 409-425.	1.1	57
613	Comprehensive Computational Analysis of GWAS Loci Identifies CCR2 as a Candidate Gene for Celiac Disease Pathogenesis. Journal of Cellular Biochemistry, 2017, 118, 2193-2207.	1.2	17
614	Mutation screening of ACKR3 and COPS8 in kidney cancer cases from the CONFIRM study. Familial Cancer, 2017, 16, 411-416.	0.9	6
615	Genome-wide association study implicates immune activation of multiple integrin genes in inflammatory bowel disease. Nature Genetics, 2017, 49, 256-261.	9.4	943
616	The primacy of NF1 loss as the driver of tumorigenesis in neurofibromatosis type 1-associated plexiform neurofibromas. Oncogene, 2017, 36, 3168-3177.	2.6	84
617	ABCA7 loss-of-function variants, expression, and neurologic disease risk. Neurology: Genetics, 2017, 3, e126.	0.9	26
618	JIMD Reports, Volume 31. JIMD Reports, 2017, , .	0.7	0
619	Discovery and functional prioritization of Parkinsonâ€™s disease candidate genes from large-scale whole exome sequencing. Genome Biology, 2017, 18, 22.	3.8	96
620	The current state of clinical interpretation of sequence variants. Current Opinion in Genetics and Development, 2017, 42, 33-39.	1.5	77

#	Article	IF	Citations
622	Absence of Hikeshi, a nuclear transporter for heat-shock protein HSP70, causes infantile hypomyelinating leukoencephalopathy. European Journal of Human Genetics, 2017, 25, 366-370.	1.4	11
623	Heterozygous Pathogenic Variant inDACT1Causes an Autosomal-Dominant Syndrome with Features Overlapping Townes-Brocks Syndrome. Human Mutation, 2017, 38, 373-377.	1.1	17
624	Structural, Functional, and Clinical Characterization of a NovelPTPN11Mutation Cluster Underlying Noonan Syndrome. Human Mutation, 2017, 38, 451-459.	1.1	39
625	Practical Approaches for Whole-Genome Sequence Analysis of Heart- and Blood-Related Traits. American Journal of Human Genetics, 2017, 100, 205-215.	2.6	50
626	POLG2 deficiency causes adultâ€onset syndromic sensory neuropathy, ataxia and parkinsonism. Annals of Clinical and Translational Neurology, 2017, 4, 4-14.	1.7	13
627	Predicting Severity of Disease-Causing Variants. Human Mutation, 2017, 38, 357-364.	1.1	33
628	FGFR1 Analyses in Four Patients with Hypogonadotropic Hypogonadism with Split-Hand/Foot Malformation: Implications for the Promoter Region. Human Mutation, 2017, 38, 503-506.	1.1	6
629	Mutation effects predicted from sequence co-variation. Nature Biotechnology, 2017, 35, 128-135.	9.4	543
630	Comprehensive Screening for Disease Risk Variants in Early-Onset Alzheimerâ€™s Disease Genes in African Americans Identifies Novel PSEN Variants. Journal of Alzheimer's Disease, 2017, 56, 1215-1222.	1.2	4
631	A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. American Journal of Human Genetics, 2017, 100, 343-351.	2.6	35
632	InterVar: Clinical Interpretation of Genetic Variants by the 2015 ACMG-AMP Guidelines. American Journal of Human Genetics, 2017, 100, 267-280.	2.6	717
633	A novel, putatively null, FGD1 variant leading to Aarskog-Scott syndrome in a family from UAE. BMC Pediatrics, 2017, 17, 31.	0.7	4
634	Molecular and clinical analysis of ALPL in a cohort of patients with suspicion of Hypophosphatasia. American Journal of Medical Genetics, Part A, 2017, 173, 601-610.	0.7	36
635	Predicting enhancer activity and variant impact using gkmâ€SVM. Human Mutation, 2017, 38, 1251-1258.	1.1	40
636	Biallelic Mutations in DNAJC12 Cause Hyperphenylalaninemia, Dystonia, and Intellectual Disability. American Journal of Human Genetics, 2017, 100, 257-266.	2.6	127
637	Predicting gene expression in massively parallel reporter assays: A comparative study. Human Mutation, 2017, 38, 1240-1250.	1.1	39
638	Molecular spectrum and differential diagnosis in patients referred with sporadic or autosomal recessive osteogenesis imperfecta. Molecular Genetics & Genomic Medicine, 2017, 5, 28-39.	0.6	37
639	Exome sequencing identifies SLC26A4, GJB2, SCARB2 and DUOX2 mutations in 2 siblings with Pendred syndrome in a Malaysian family. Orphanet Journal of Rare Diseases, 2017, 12, 40.	1.2	7

#	Article	IF	Citations
640	PONâ€P and PONâ€P2 predictor performance in CAGI challenges: Lessons learned. Human Mutation, 2017, 38, 1085-1091.	1.1	5
641	A novel <scp>DNAJB6</scp> mutation causes dominantly inherited distalâ€onset myopathy and compromises <scp>DNAJB6</scp> function. Clinical Genetics, 2017, 92, 150-157.	1.0	16
642	Whole genome sequencing of one complex pedigree illustrates challenges with genomic medicine. BMC Medical Genomics, 2017, 10, 10.	0.7	15
643	Genetic variants in the peripheral auditory system significantly affect adult cochlear implant performance. Hearing Research, 2017, 348, 138-142.	0.9	68
644	GAVIN: Gene-Aware Variant INterpretation for medical sequencing. Genome Biology, 2017, 18, 6.	3.8	55
645	A girl with developmental delay, ataxia, cranial nerve palsies, severe respiratory problems in infancyâ€"Expanding NDST1 syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 712-715.	0.7	14
646	Exome analysis of Smithâ€"Magenis-like syndrome cohort identifies de novo likely pathogenic variants. Human Genetics, 2017, 136, 409-420.	1.8	22
647	From exomes to genomes: challenges and solutions in population-based genetic association studies. European Journal of Human Genetics, 2017, 25, 395-396.	1.4	4
648	Identification of Cadherin 2 (CDH2) Mutations in Arrhythmogenic Right Ventricular Cardiomyopathy. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	123
649	Rare deleterious variants in GRHL3 are associated with human spina bifida. Human Mutation, 2017, 38, 716-724.	1.1	28
650	Impacts of Neanderthal-Introgressed Sequences on the Landscape of Human Gene Expression. Cell, 2017, 168, 916-927.e12.	13.5	136
651	Human Adaptive Immunity Rescues an Inborn Error of Innate Immunity. Cell, 2017, 168, 789-800.e10.	13.5	68
652	The performance of deleteriousness prediction scores for rare non-protein-changing single nucleotide variants in human genes. Journal of Medical Genetics, 2017, 54, 134-144.	1.5	16
653	Accurate eQTL prioritization with an ensembleâ€based framework. Human Mutation, 2017, 38, 1259-1265.	1.1	15
654	A de novo splice site mutation in CASK causes FG syndromeâ€4 and congenital nystagmus. American Journal of Medical Genetics, Part A, 2017, 173, 611-617.	0.7	11
655	Putative TMPRSS3/GJB2 digenic inheritance of hearing loss detected by targeted resequencing. Molecular and Cellular Probes, 2017, 33, 24-27.	0.9	8
656	Common genetic variation and risk of gallbladder cancer in India: a case-control genome-wide association study. Lancet Oncology, The, 2017, 18, 535-544.	5.1	69
657	Possible role of rare variants in Trace amine associated receptor 1 in schizophrenia. Schizophrenia Research, 2017, 189, 190-195.	1.1	40

#	Article	IF	Citations
658	Whole genome sequencing resource identifies 18 new candidate genes for autism spectrum disorder. Nature Neuroscience, 2017, 20, 602-611.	7.1	691
659	Identification of novel <scp>SNORD118</scp> mutations in seven patients with leukoencephalopathy with brain calcifications and cysts. Clinical Genetics, 2017, 92, 180-187.	1.0	28
660	Molecular genetic findings and clinical correlations in 100 patients with Joubert syndrome and related disorders prospectively evaluated at a single center. Genetics in Medicine, 2017, 19, 875-882.	1.1	100
661	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. Nature Genetics, 2017, 49, 515-526.	9.4	443
662	Mutations in INPP5K Cause a Form of Congenital Muscular Dystrophy Overlapping Marinesco-SjÃ¶gren Syndrome and Dystroglycanopathy. American Journal of Human Genetics, 2017, 100, 537-545.	2.6	67
663	High diagnostic yield of syndromic intellectual disability by targeted next-generation sequencing. Journal of Medical Genetics, 2017, 54, 87-92.	1.5	93
664	Opportunities and challenges of whole-genome and -exome sequencing. BMC Genetics, 2017, 18, 14.	2.7	160
665	Host genetic variation in mucosal immunity pathways influences the upper airway microbiome. Microbiome, 2017, 5, 16.	4.9	61
666	Geneâ€based segregation method for identifying rare variants in familyâ€based sequencing studies. Genetic Epidemiology, 2017, 41, 309-319.	0.6	14
667	Purinergic receptorsP2RX4andP2RX7in familial multiple sclerosis. Human Mutation, 2017, 38, 736-744.	1.1	46
668	Postmortem genetic testing should be recommended in sudden cardiac death cases due to thoracic aortic dissection. International Journal of Legal Medicine, 2017, 131, 1211-1219.	1.2	13
669	Population- and individual-specific regulatory variation in Sardinia. Nature Genetics, 2017, 49, 700-707.	9.4	38
670	Genome-wide association analysis for chronic venous disease identifies EFEMP1 and KCNH8 as susceptibility loci. Scientific Reports, 2017, 7, 45652.	1.6	48
671	Biotinâ€thiamine responsive basal ganglia disease: Identification of a pyruvate peak on brain spectroscopy, novel mutation in SLC19A3, and calculation of prevalence based on allele frequencies from aggregated nextâ€generation sequencing data. American Journal of Medical Genetics, Part A, 2017, 173, 1502-1513.	0.7	15
672	Germline Mutations in CDH23, Encoding Cadherin-Related 23, Are Associated with Both Familial and Sporadic Pituitary Adenomas. American Journal of Human Genetics, 2017, 100, 817-823.	2.6	57
673	Nkx2-5 and Sarcospan genetically interact in the development of the muscular ventricular septum of the heart. Scientific Reports, 2017, 7, 46438.	1.6	6
674	Leveraging cell type specific regulatory regions to detect SNPs associated with tissue factor pathway inhibitor plasma levels. Genetic Epidemiology, 2017, 41, 455-466.	0.6	1
675	Computational Analysis of Breast Cancer GWAS Loci Identifies the Putative Deleterious Effect of STXBP4 and ZNF404 Gene Variants. Journal of Cellular Biochemistry, 2017, 118, 4296-4307.	1.2	11

#	Article	IF	Citations
676	Gene and Variant Annotation for Mendelian Disorders in the Era of Advanced Sequencing Technologies. Annual Review of Genomics and Human Genetics, 2017, 18, 229-256.	2.5	37
677	Behavioural characterization of AnkyrinG deficient mice, a model for ANK3 related disorders. Behavioural Brain Research, 2017, 328, 218-226.	1.2	16
678	Utility of Post-Mortem Genetic Testing in Cases of Sudden Arrhythmic Death Syndrome. Journal of the American College of Cardiology, 2017, 69, 2134-2145.	1.2	219
679	Selection in Europeans on Fatty Acid Desaturases Associated with Dietary Changes. Molecular Biology and Evolution, 2017, 34, 1307-1318.	3.5	90
680	A case of splenomegaly in CBL syndrome. European Journal of Medical Genetics, 2017, 60, 374-379.	0.7	10
681	Genetic Identification in Early Onset Parkinsonism among Norwegian Patients. Movement Disorders Clinical Practice, 2017, 4, 499-508.	0.8	25
682	Enteropathy-associated T cell lymphoma subtypes are characterized by loss of function of SETD2. Journal of Experimental Medicine, 2017, 214, 1371-1386.	4.2	144
683	Combating mutations in genetic disease and drug resistance: understanding molecular mechanisms to guide drug design. Expert Opinion on Drug Discovery, 2017, 12, 553-563.	2.5	26
684	mTCTScan: a comprehensive platform for annotation and prioritization of mutations affecting drug sensitivity in cancers. Nucleic Acids Research, 2017, 45, W215-W221.	6.5	12
685	De novo REEP2 missense mutation in pure hereditary spastic paraplegia. Annals of Clinical and Translational Neurology, 2017, 4, 347-350.	1.7	9
686	Determination of disease phenotypes and pathogenic variants from exome sequence data in the CAGI 4 gene panel challenge. Human Mutation, 2017, 38, 1201-1216.	1.1	5
687	Limited influence of germline genetic variation on all-cause mortality in women with early onset breast cancer: evidence from gene-based tests, single-marker regression, and whole-genome prediction. Breast Cancer Research and Treatment, 2017, 164, 707-717.	1.1	4
688	Targeted sequencing identifies novel variants involved in autosomal recessive hereditary hearing loss in Qatari families. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2017, 800-802, 29-36.	0.4	23
689	A Novel Kleefstra Syndrome-associated Variant That Affects the Conserved TPLX Motif within the Ankyrin Repeat of EHMT1 Leads to Abnormal Protein Folding. Journal of Biological Chemistry, 2017, 292, 3866-3876.	1.6	18
690	Pathogenic Anti-MÃ¼llerian Hormone Variants in Polycystic Ovary Syndrome. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 2862-2872.	1.8	80
691	Germline SAMD9 mutation in siblings with monosomy 7 and myelodysplastic syndrome. Leukemia, 2017, 31, 1827-1830.	3.3	58
692	Rare NOX3 Variants Confer Susceptibility to Agranulocytosis During Thyrostatic Treatment of Graves' Disease. Clinical Pharmacology and Therapeutics, 2017, 102, 1017-1024.	2.3	12
693	A pathway-centric approach to rare variant association analysis. European Journal of Human Genetics, 2017, 25, 123-129.	1.4	13

#	Article	IF	Citations
694	Whole-genome landscapes of major melanoma subtypes. Nature, 2017, 545, 175-180.	13.7	1,068
695	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. Brain, 2017, 140, 1561-1578.	3.7	85
696	Genome-Wide Association Studies and Meta-Analyses for Congenital Heart Defects. Circulation: Cardiovascular Genetics, 2017, 10, e001449.	5.1	47
697	Identification of Somatic Mutations in Primary Cutaneous Diffuse Large B-Cell Lymphoma, Leg Type by Massive Parallel Sequencing. Journal of Investigative Dermatology, 2017, 137, 1984-1994.	0.3	93
698	Rare missense mutations in P2RY11 in narcolepsy with cataplexy. Brain, 2017, 140, 1657-1668.	3.7	27
699	Severe congenital microcephaly with AP4M1 mutation, a case report. BMC Medical Genetics, 2017, 18, 48.	2.1	6
700	Deleterious ABCA7 mutations and transcript rescue mechanisms in early onset Alzheimerâ€™s disease. Acta Neuropathologica, 2017, 134, 475-487.	3.9	53
701	Functional Investigations of HNF1A Identify Rare Variants as Risk Factors for Type 2 Diabetes in the General Population. Diabetes, 2017, 66, 335-346.	0.3	54
702	PCSK9 genetic variants and risk of type 2 diabetes: a mendelian randomisation study. Lancet Diabetes and Endocrinology,the, 2017, 5, 97-105.	5.5	298
703	Rare Genetic Variant in SORL1 May Increase Penetrance of Alzheimerâ€™s Disease in a Family with Several Generations of APOE-É›4 Homozygosity. Journal of Alzheimer's Disease, 2017, 56, 63-74.	1.2	32
704	Variant in the X-chromosome spliceosomal gene GPKOW causes male-lethal microcephaly with intrauterine growth restriction. European Journal of Human Genetics, 2017, 25, 1078-1082.	1.4	10
705	Pheno4J: a gene to phenotype graph database. Bioinformatics, 2017, 33, 3317-3319.	1.8	9
706	Mutations in ARMC9, which Encodes a Basal Body Protein, Cause Joubert Syndrome in Humans and Ciliopathy Phenotypes in Zebrafish. American Journal of Human Genetics, 2017, 101, 23-36.	2.6	74
707	Genetic Approaches to Understanding Psychiatric Disease. Neurotherapeutics, 2017, 14, 564-581.	2.1	6
708	The phenotypic spectrum of ARHGEF9 includes intellectual disability, focal epilepsy and febrile seizures. Journal of Neurology, 2017, 264, 1421-1425.	1.8	20
709	HIPred: an integrative approach to predicting haploinsufficient genes. Bioinformatics, 2017, 33, 1751-1757.	1.8	36
710	Expression quantitative trait loci for PI3K/AKT pathway. Medicine (United States), 2017, 96, e5817.	0.4	11
711	Epilepsy-associated GRIN2A mutations reduce NMDA receptor trafficking and agonist potency â€“ molecular profiling and functional rescue. Scientific Reports, 2017, 7, 66.	1.6	74

#	Article	IF	Citations
712	Common sequence variants affect molecular function more than rare variants?. Scientific Reports, 2017, 7, 1608.	1.6	20
713	NeuroChip, an updated version of the NeuroX genotyping platform to rapidly screen for variants associated with neurological diseases. Neurobiology of Aging, 2017, 57, 247.e9-247.e13.	1.5	108
714	Variants in TTC25 affect autistic trait in patients with autism spectrum disorder and general population. European Journal of Human Genetics, 2017, 25, 982-987.	1.4	5
715	CAGI4 SickKids clinical genomes challenge: A pipeline for identifying pathogenic variants. Human Mutation, 2017, 38, 1169-1181.	1.1	11
717	GeMSTONE: orchestrated prioritization of human germline mutations in the cloud. Nucleic Acids Research, 2017, 45, W207-W214.	6.5	2
718	Bioinformatics Tools for Genome-Wide Epigenetic Research. Advances in Experimental Medicine and Biology, 2017, 978, 489-512.	0.8	43
719	KMT2B rare missense variants in generalized dystonia. Movement Disorders, 2017, 32, 1087-1091.	2.2	42
720	CAGI4 Crohn's exome challenge: Marker SNP versus exome variant models for assigning risk of Crohn disease. Human Mutation, 2017, 38, 1225-1234.	1.1	15
721	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. Nature Neuroscience, 2017, 20, 1043-1051.	7.1	152
722	MCM3AP in recessive Charcot-Marie-Tooth neuropathy and mild intellectual disability. Brain, 2017, 140, 2093-2103.	3.7	31
723	Metastatic triple-negative breast cancer patient with TP53 tumor mutation experienced 11 months progression-free survival on bortezomib monotherapy without adverse events after ending standard treatments with grade 3 adverse events. Journal of Physical Education and Sports Management, 2017, 3, a001677.	0.5	14
724	Identification of 8 Novel Mutations in Nephrogenesis-Related Genes in Chinese Han Patients with Unilateral Renal Agenesis. American Journal of Nephrology, 2017, 46, 55-63.	1.4	11
725	Regulatory element-based prediction identifies new susceptibility regulatory variants for osteoporosis. Human Genetics, 2017, 136, 963-974.	1.8	11
726	Imbalance-Aware Machine Learning for Predicting Rare and Common Disease-Associated Non-Coding Variants. Scientific Reports, 2017, 7, 2959.	1.6	63
727	Ensemble variant interpretation methods to predict enzyme activity and assign pathogenicity in the CAGI4 NAGLU (Human Nâ€acetylâ€glucosaminidase) and UBE2I (Human SUMOâ€ligase) challenges. Human Mutation, 2017, 38, 1109-1122.	1.1	14
728	Application of pharmacogenomics to investigate adverse drug reactions to the disease-modifying treatments for multiple sclerosis: a caseâ€“control study protocol for dimethyl fumarate-induced lymphopenia. BMJ Open, 2017, 7, e016276.	0.8	2
729	Challenges and progress in interpretation of non-coding genetic variants associated with human disease. Experimental Biology and Medicine, 2017, 242, 1325-1334.	1.1	44
730	Genetic Testing in the Evaluation of Unexplained Cardiac Arrest. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	76

#	Article	IF	Citations
731	Semi-automated cancer genome analysis using high-performance computing. Human Mutation, 2017, 38, 1325-1335.	1.1	9
732	Genome-wide association meta-analysis of 78,308 individuals identifies new loci and genes influencing human intelligence. Nature Genetics, 2017, 49, 1107-1112.	9.4	425
733	BACH2 immunodeficiency illustrates an association between super-enhancers and haploinsufficiency. Nature Immunology, 2017, 18, 813-823.	7.0	113
734	HGVA: the Human Genome Variation Archive. Nucleic Acids Research, 2017, 45, W189-W194.	6.5	6
735	Nextâ€generation sequencing of a family with a high penetrance of monoclonal gammopathies for the identification of candidate risk alleles. Cancer, 2017, 123, 3701-3708.	2.0	12
736	Objective assessment of the evolutionary action equation for the fitness effect of missense mutations across CAGIâ€blinded contests. Human Mutation, 2017, 38, 1072-1084.	1.1	26
737	Canine Brachycephaly Is Associated with a Retrotransposon-Mediated Missplicing of SMOC2. Current Biology, 2017, 27, 1573-1584.e6.	1.8	80
738	Protein-altering and regulatory genetic variants near GATA4 implicated in bicuspid aortic valve. Nature Communications, 2017, 8, 15481.	5.8	90
739	Characterization of pathogenic SORL1 genetic variants for association with Alzheimerâ€™s disease: a clinical interpretation strategy. European Journal of Human Genetics, 2017, 25, 973-981.	1.4	102
740	ACSS2 gene variant associated with cleft lip and palate in two independent Hispanic populations. Laryngoscope, 2017, 127, E336-E339.	1.1	2
741	PMut: a web-based tool for the annotation of pathological variants on proteins, 2017 update. Nucleic Acids Research, 2017, 45, W222-W228.	6.5	184
742	Recurrent elevated liver transaminases and acute liver failure in two siblings with novel bi-allelic mutations of NBAS. European Journal of Medical Genetics, 2017, 60, 426-432.	0.7	31
743	Genetic subclone architecture of tumor clone-initiating cells in colorectal cancer. Journal of Experimental Medicine, 2017, 214, 2073-2088.	4.2	30
744	Genomic diagnosis for children with intellectual disability and/or developmental delay. Genome Medicine, 2017, 9, 43.	3.6	188
745	Identification of a rare LAMB4 variant associated with familial diverticulitis through exome sequencing. Human Molecular Genetics, 2017, 26, 3212-3220.	1.4	15
746	Colonic Phenotypes Are Associated with Poorer Response to Anti-TNF Therapies in Patients with IBD. Inflammatory Bowel Diseases, 2017, 23, 1382-1393.	0.9	22
747	Supporting precision medicine by data mining across multi-disciplines: an integrative approach for generating comprehensive linkages between single nucleotide variants (SNVs) and drug-binding sites. Bioinformatics, 2017, 33, 1621-1629.	1.8	11
748	Accurately annotate compound effects of genetic variants using a context-sensitive framework. Nucleic Acids Research, 2017, 45, e82-e82.	6.5	9

#	Article	IF	Citations
749	Homozygous p.(Glu87Lys) variant in ISCA1 is associated with a multiple mitochondrial dysfunctions syndrome. Journal of Human Genetics, 2017, 62, 723-727.	1.1	53
750	Development and Validation of Clinical Whole-Exome and Whole-Genome Sequencing for Detection of Germline Variants in Inherited Disease. Archives of Pathology and Laboratory Medicine, 2017, 141, 798-805.	1.2	46
751	Unravelling the genetics of inherited retinal dystrophies: Past, present and future. Progress in Retinal and Eye Research, 2017, 59, 53-96.	7.3	85
752	The impact of structural variation on human gene expression. Nature Genetics, 2017, 49, 692-699.	9.4	334
753	A recurrent WARS mutation is a novel cause of autosomal dominant distal hereditary motor neuropathy. Brain, 2017, 140, 1252-1266.	3.7	75
754	Compound heterozygous mutations in the gene PIGP are associated with early infantile epileptic encephalopathy. Human Molecular Genetics, 2017, 26, 1706-1715.	1.4	39
755	Populationâ€Specific Resequencing Associates the ATPâ€Binding Cassette Subfamily C Member 4 Gene With Gout in New Zealand MÄori and Pacific Men. Arthritis and Rheumatology, 2017, 69, 1461-1469.	2.9	46
756	Data resources for the identification and interpretation of actionable mutations by clinicians. Annals of Oncology, 2017, 28, 946-957.	0.6	20
757	TLR3 Mutations in Adult Patients With Herpes Simplex Virus and Varicella-Zoster Virus Encephalitis. Journal of Infectious Diseases, 2017, 215, 1430-1434.	1.9	53
758	Large-Scale Profiling Reveals the Influence of Genetic Variation on Gene Expression in Human Induced Pluripotent Stem Cells. Cell Stem Cell, 2017, 20, 533-546.e7.	5.2	157
759	Malignant hyperthermia susceptibility in patients with exertional rhabdomyolysis: a retrospective cohort study and updated systematic review. Canadian Journal of Anaesthesia, 2017, 64, 736-743.	0.7	31
760	PUF60 variants cause a syndrome of ID, short stature, microcephaly, coloboma, craniofacial, cardiac, renal and spinal features. European Journal of Human Genetics, 2017, 25, 552-559.	1.4	42
761	Whole exome sequencing with genomic triangulation implicatesCDH2-encoded N-cadherin as a novel pathogenic substrate for arrhythmogenic cardiomyopathy. Congenital Heart Disease, 2017, 12, 226-235.	0.0	46
762	Common genetic etiology between â€œmultiple sclerosis-likeâ€•single-gene disorders and familial multiple sclerosis. Human Genetics, 2017, 136, 705-714.	1.8	12
763	Functional variomics and network perturbation: connecting genotype to phenotype in cancer. Nature Reviews Genetics, 2017, 18, 395-410.	7.7	84
764	Autosomal Recessive Cardiomyopathy Presenting as Acute Myocarditis. Journal of the American College of Cardiology, 2017, 69, 1653-1665.	1.2	94
765	Predicting the impact of non-coding variants on DNA methylation. Nucleic Acids Research, 2017, 45, e99-e99.	6.5	81
766	Molecular basis of familial hypercholesterolemia. Current Opinion in Cardiology, 2017, 32, 262-266.	0.8	25

#	Article	IF	Citations
767	AMMECR1: a single point mutation causes developmental delay, midface hypoplasia and elliptocytosis. Journal of Medical Genetics, 2017, 54, 269-277.	1.5	12
768	Deep intronic variants introduce DMD pseudoexon in patient with muscular dystrophy. Neuromuscular Disorders, 2017, 27, 631-634.	0.3	17
769	Mortality in Joubert syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 1237-1242.	0.7	22
770	Screening of the Filamin C Gene in a Large Cohort of Hypertrophic Cardiomyopathy Patients. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	68
771	Recurrent epigenetic silencing of the PTPRD tumor suppressor in laryngeal squamous cell carcinoma. Tumor Biology, 2017, 39, 101042831769142.	0.8	12
772	A comprehensive analysis of cancer-driving mutations and genes in kidney cancer. Oncology Letters, 2017, 13, 2151-2160.	0.8	4
773	Genome-wide meta-analysis identifies novel loci of plaque burden in carotid artery. Atherosclerosis, 2017, 259, 32-40.	0.4	33
774	Fast, scalable prediction of deleterious noncoding variants from functional and population genomic data. Nature Genetics, 2017, 49, 618-624.	9.4	299
775	Identification of Elongated Primary Cilia with Impaired Mechanotransduction in Idiopathic Scoliosis Patients. Scientific Reports, 2017, 7, 44260.	1.6	44
776	Loss of LMOD1 impairs smooth muscle cytocontractility and causes megacystis microcolon intestinal hypoperistalsis syndrome in humans and mice. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E2739-E2747.	3.3	97
777	A truncating mutation in CEP55 is the likely cause of MARCH, a novel syndrome affecting neuronal mitosis. Journal of Medical Genetics, 2017, 54, 490-501.	1.5	45
778	cepip: context-dependent epigenomic weighting for prioritization of regulatory variants and disease-associated genes. Genome Biology, 2017, 18, 52.	3.8	33
779	Alternating Hemiplegia of Childhood as a New Presentation of Adenylate Cyclase 5-Mutation-Associated Disease: A Report of Two Cases. Journal of Pediatrics, 2017, 181, 306-308.e1.	0.9	24
780	Mining the Unknown: Assigning Function to Noncoding Single Nucleotide Polymorphisms. Trends in Genetics, 2017, 33, 34-45.	2.9	81
781	PERCH: A Unified Framework for Disease Gene Prioritization. Human Mutation, 2017, 38, 243-251.	1.1	119
782	Combined immunodeficiency and Epstein-Barr virusâ€“induced B cell malignancy in humans with inherited CD70 deficiency. Journal of Experimental Medicine, 2017, 214, 91-106.	4.2	134
783	De Novo Mutations in EBF3 Cause a Neurodevelopmental Syndrome. American Journal of Human Genetics, 2017, 100, 138-150.	2.6	52
784	Germline and somatic DICER1 mutations in familial and sporadic liver tumors. Journal of Hepatology, 2017, 66, 734-742.	1.8	31

#	Article	IF	Citations
785	Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. American Journal of Human Genetics, 2017, 100, 75-90.	2.6	343
786	Evaluating the role of TMEM230 variants in Parkinson's disease. Parkinsonism and Related Disorders, 2017, 35, 100-101.	1.1	15
787	Alanine-scanning mutagenesis of human signal transducer and activator of transcription 1 to estimate loss- or gain-of-function variants. Journal of Allergy and Clinical Immunology, 2017, 140, 232-241.	1.5	43
788	A Genome-wide Association Study Identifies Risk Alleles in Plasminogen and P4HA2 Associated with Giant Cell Arteritis. American Journal of Human Genetics, 2017, 100, 64-74.	2.6	78
789	Mutations in <scp>DYNC2H1</scp>, the cytoplasmic dynein 2, heavy chain 1 motor protein gene, cause shortâ€rib polydactyly type I, Saldinoâ€“Noonan type. Clinical Genetics, 2017, 92, 158-165.	1.0	21
790	The Rare-Variant Generalized Disequilibrium Test for Association Analysis of Nuclear and Extended Pedigrees with Application to Alzheimer Disease WGS Data. American Journal of Human Genetics, 2017, 100, 193-204.	2.6	26
791	denovo-db: a compendium of humande novovariants. Nucleic Acids Research, 2017, 45, D804-D811.	6.5	173
792	Enrichment of deleterious variants of mitochondrial DNA polymerase gene (POLG1) in bipolar disorder. Psychiatry and Clinical Neurosciences, 2017, 71, 518-529.	1.0	29
793	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. Nature Genetics, 2017, 49, 223-237.	9.4	186
794	A novel class of somatic mutations in blood detected preferentially in CD8 + cells. Clinical Immunology, 2017, 175, 75-81.	1.4	35
795	Mutations in two large pedigrees highlight the role of ZNF711 in X-linked intellectual disability. Gene, 2017, 605, 92-98.	1.0	26
796	Standards and Guidelines for the Interpretation and Reporting of Sequence Variants in Cancer. Journal of Molecular Diagnostics, 2017, 19, 4-23.	1.2	1,267
797	Dawning of the age of genomics for platelet granule disorders: improving insight, diagnosis and management. British Journal of Haematology, 2017, 176, 705-720.	1.2	22
798	Analysis commons, a team approach to discovery in a big-data environment for genetic epidemiology. Nature Genetics, 2017, 49, 1560-1563.	9.4	93
799	Racial disparities in patient survival and tumor mutation burden, and the association between tumor mutation burden and cancer incidence rate. Scientific Reports, 2017, 7, 13639.	1.6	37
800	MAPPIN: a method for annotating, predicting pathogenicity and mode of inheritance for nonsynonymous variants. Nucleic Acids Research, 2017, 45, 10393-10402.	6.5	15
801	Exome-wide Association Study Identifies GREB1L Mutations in Congenital Kidney Malformations. American Journal of Human Genetics, 2017, 101, 789-802.	2.6	63
802	Natural Selection on Genes Related to Cardiovascular Health in High-Altitude Adapted Andeans. American Journal of Human Genetics, 2017, 101, 752-767.	2.6	99

#	Article	IF	Citations
803	Phenotypic Characterization of EIF2AK4 Mutation Carriers in a Large Cohort of Patients Diagnosed Clinically With Pulmonary Arterial Hypertension. Circulation, 2017, 136, 2022-2033.	1.6	111
804	Genomic Triangulation and Coverage Analysis in Whole-Exome Sequencingâ€“Based Molecular Autopsies. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	14
805	The impact of rare variation on gene expression across tissues. Nature, 2017, 550, 239-243.	13.7	229
806	Whole exome sequencing in inborn errors of immunity: use the power but mind the limits. Current Opinion in Allergy and Clinical Immunology, 2017, 17, 421-430.	1.1	12
807	Rapid Targeted Genomics in Critically Ill Newborns. Pediatrics, 2017, 140, .	1.0	99
808	Functional analysis of novelDEAF1variants identified through clinical exome sequencing expandsDEAF1-associated neurodevelopmental disorder (DAND) phenotype. Human Mutation, 2017, 38, 1774-1785.	1.1	20
810	Overexpressed somatic alleles are enriched in functional elements in Breast Cancer. Scientific Reports, 2017, 7, 8287.	1.6	3
811	Genomic Patterns of De Novo Mutation in Simplex Autism. Cell, 2017, 171, 710-722.e12.	13.5	308
812	Evidence of Early-Stage Selection on EPAS1 and GPR126 Genes in Andean High Altitude Populations. Scientific Reports, 2017, 7, 13042.	1.6	29
813	Protein interaction perturbation profiling at amino-acid resolution. Nature Methods, 2017, 14, 1213-1221.	9.0	36
814	Signatures of positive selection reveal a universal role of chromatin modifiers as cancer driver genes. Scientific Reports, 2017, 7, 13124.	1.6	20
815	Analysis and Annotation of Wholeâ€Genome or Wholeâ€Exome Sequencing Derived Variants for Clinical Diagnosis. Current Protocols in Human Genetics, 2017, 95, 9.24.1-9.24.28.	3.5	20
816	De novo mutations of TUBA3D are associated with keratoconus. Scientific Reports, 2017, 7, 13570.	1.6	20
817	Association analysis identifies 65 new breast cancer risk loci. Nature, 2017, 551, 92-94.	13.7	1,099
818	Evaluating phenotype-driven approaches for genetic diagnoses from exomes in a clinical setting. Scientific Reports, 2017, 7, 13509.	1.6	26
819	Clinical genetics and outcome of left ventricular non-compaction cardiomyopathy. European Heart Journal, 2017, 38, 3449-3460.	1.0	168
820	De novo, deleterious sequence variants that alter the transcriptional activity of the homeoprotein PBX1 are associated with intellectual disability and pleiotropic developmental defects. Human Molecular Genetics, 2017, 26, 4849-4860.	1.4	42
821	The GBA variant E326K is associated with Parkinson's disease and explains a genome-wide association signal. Neuroscience Letters, 2017, 658, 48-52.	1.0	57

#	Article	IF	Citations
822	Unified Sequence-Based Association Tests Allowing for Multiple Functional Annotations and Meta-analysis of Noncoding Variation in Metabochip Data. American Journal of Human Genetics, 2017, 101, 340-352.	2.6	45
823	A Scalable Bayesian Method for Integrating Functional Information in Genome-wide Association Studies. American Journal of Human Genetics, 2017, 101, 404-416.	2.6	63
824	Panel sequencing of 264 candidate susceptibility genes and segregation analysis in a cohort of non-BRCA1, non-BRCA2 breast cancer families. Breast Cancer Research and Treatment, 2017, 166, 937-949.	1.1	16
825	FIRE: functional inference of genetic variants that regulate gene expression. Bioinformatics, 2017, 33, 3895-3901.	1.8	30
826	Assessing predictions of fitness effects of missense mutations in SUMOâ€conjugating enzyme UBE2I. Human Mutation, 2017, 38, 1051-1063.	1.1	12
827	The Use of Variant Maps to Explore Domain-Specific Mutations of FGFR1. Journal of Dental Research, 2017, 96, 1339-1345.	2.5	7
828	A clinically driven variant prioritization framework outperforms purely computational approaches for the diagnostic analysis of singleton WES data. European Journal of Human Genetics, 2017, 25, 1268-1272.	1.4	24
829	Molecular diversity of combined and complex dystonia: insights from diagnostic exome sequencing. Neurogenetics, 2017, 18, 195-205.	0.7	37
830	Ultra-rare mutations in SRCAP segregate in Caribbean Hispanic families with Alzheimer disease. Neurology: Genetics, 2017, 3, e178.	0.9	8
831	Characterizing clinically relevant natural variants of GPCRs using computational approaches. Methods in Cell Biology, 2017, 142, 187-204.	0.5	7
832	Identification of ASAH1 as a susceptibility gene for familial keloids. European Journal of Human Genetics, 2017, 25, 1155-1161.	1.4	19
833	Mind the Gap: Current Challenges and Future State of Heart Failure Care. Canadian Journal of Cardiology, 2017, 33, 1434-1449.	0.8	19
834	Discovery of Variants Underlying Host Susceptibility to Virus Infection Using Whole-Exome Sequencing. Methods in Molecular Biology, 2017, 1656, 209-227.	0.4	0
835	Prevalence of common hereditary risk factors for thrombophilia in Somalia and identification of a novel Gln544Arg mutation in coagulation factor V. Journal of Thrombosis and Thrombolysis, 2017, 44, 536-543.	1.0	8
836	A disease-associated mutation in the adhesion GPCR BAI2 (ADGRB2) increases receptor signaling activity. Human Mutation, 2017, 38, 1751-1760.	1.1	24
837	Exome sequences of multiplex, multigenerational families reveal schizophrenia risk loci with potential implications for neurocognitive performance. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2017, 174, 817-827.	1.1	8
838	Sasquatch: predicting the impact of regulatory SNPs on transcription factor binding from cell- and tissue-specific DNase footprints. Genome Research, 2017, 27, 1730-1742.	2.4	33
839	Targeted Gene Panel Sequencing for Early-onset Inflammatory Bowel Disease and Chronic Diarrhea. Inflammatory Bowel Diseases, 2017, 23, 2109-2120.	0.9	33

#	Article	IF	Citations
840	Annotating pathogenic non-coding variants in genic regions. Nature Communications, 2017, 8, 236.	5.8	122
841	Susceptible genes and disease mechanisms identified in frontotemporal dementia and frontotemporal dementia with Amyotrophic Lateral Sclerosis by DNA-methylation and GWAS. Scientific Reports, 2017, 7, 8899.	1.6	30
842	CScape: a tool for predicting oncogenic single-point mutations in the cancer genome. Scientific Reports, 2017, 7, 11597.	1.6	52
843	Encephalopathy caused by novel mutations in the CMPâ€sialic acid transporter, SLC35A1. American Journal of Medical Genetics, Part A, 2017, 173, 2906-2911.	0.7	26
844	Genomic landscape of human erythroleukemia K562 cell line, as determined by next-generation sequencing and cytogenetics. Acta Haematologica Polonica, 2017, 48, 343-349.	0.1	3
845	Optimizing genomic medicine in epilepsy through a gene-customized approach to missense variant interpretation. Genome Research, 2017, 27, 1715-1729.	2.4	150
846	Genetic regulatory effects modified by immune activation contribute to autoimmune disease associations. Nature Communications, 2017, 8, 266.	5.8	157
847	Using ALoFT to determine the impact of putative loss-of-function variants in protein-coding genes. Nature Communications, 2017, 8, 382.	5.8	40
848	Polygenic determinants in extremes of high-density lipoprotein cholesterol. Journal of Lipid Research, 2017, 58, 2162-2170.	2.0	49
849	The correlation between CRB1 variants and the clinical severity of Brazilian patients with different inherited retinal dystrophy phenotypes. Scientific Reports, 2017, 7, 8654.	1.6	17
850	Novel mechanisms of PIEZO1 dysfunction in hereditary xerocytosis. Blood, 2017, 130, 1845-1856.	0.6	101
851	Refining genotypeâ€“phenotype correlation in AlstrÃ¶m syndrome through study of primary human fibroblasts. Molecular Genetics & Genomic Medicine, 2017, 5, 390-404.	0.6	22
852	Survival among children with â€œLethalâ€•congenital contracture syndrome 11 caused by novel mutations in the gliomedin gene (GLDN). Human Mutation, 2017, 38, 1477-1484.	1.1	19
853	The Role of Noncoding Genetic Variation in Isolated Orofacial Clefts. Journal of Dental Research, 2017, 96, 1238-1247.	2.5	20
854	A Temporal Perspective on the Interplay of Demography and Selection on Deleterious Variation in Humans. G3: Genes, Genomes, Genetics, 2017, 7, 1027-1037.	0.8	14
855	A Zoom-Focus algorithm (ZFA) to locate the optimal testing region for rare variant association tests. Bioinformatics, 2017, 33, 2330-2336.	1.8	4
856	CRISPR/Cas9-Mediated Scanning for Regulatory Elements Required for HPRT1 Expression via Thousands of Large, Programmed Genomic Deletions. American Journal of Human Genetics, 2017, 101, 192-205.	2.6	133
857	Association analyses based on false discovery rate implicate new loci for coronary artery disease. Nature Genetics, 2017, 49, 1385-1391.	9.4	571

#	Article	IF	Citations
858	Rates, distribution and implications of postzygotic mosaic mutations in autism spectrum disorder. Nature Neuroscience, 2017, 20, 1217-1224.	7.1	212
859	<scp>MED</scp> resulting from recessively inherited mutations in the gene encoding calciumâ€activated nucleotidase <scp>CANT1</scp>. American Journal of Medical Genetics, Part A, 2017, 173, 2415-2421.	0.7	21
860	A TUBB6 mutation is associated with autosomal dominant non-progressive congenital facial palsy, bilateral ptosis and velopharyngeal dysfunction. Human Molecular Genetics, 2017, 26, 4055-4066.	1.4	13
861	The prevalence of DICER1 pathogenic variation in population databases. International Journal of Cancer, 2017, 141, 2030-2036.	2.3	75
862	A neurogenetic model for the study of schizophrenia spectrum disorders: the International 22q11.2 Deletion Syndrome Brain Behavior Consortium. Molecular Psychiatry, 2017, 22, 1664-1672.	4.1	65
863	Leveraging multiple genomic data to prioritize disease-causing indels from exome sequencing data. Scientific Reports, 2017, 7, 1804.	1.6	3
864	A population-specific reference panel empowers genetic studies of Anabaptist populations. Scientific Reports, 2017, 7, 6079.	1.6	16
865	Evidence of selection on splicing-associated loci in human populations and relevance to disease loci mapping. Scientific Reports, 2017, 7, 5980.	1.6	10
866	Whole exome sequencing reveals inherited and de novo variants in autism spectrum disorder: a trio study from Saudi families. Scientific Reports, 2017, 7, 5679.	1.6	70
867	Genetic variants including markers from the exome chip and metabolite traits of type 2 diabetes. Scientific Reports, 2017, 7, 6037.	1.6	12
868	Analysis of somatic mutations across the kinome reveals loss-of-function mutations in multiple cancer types. Scientific Reports, 2017, 7, 6418.	1.6	10
869	Integrative whole-genome sequence analysis reveals roles of regulatory mutations in BCL6 and BCL2 in follicular lymphoma. Scientific Reports, 2017, 7, 7040.	1.6	18
870	Expanding the phenotype of DST â€related disorder: A case report suggesting a genotype/phenotype correlation. American Journal of Medical Genetics, Part A, 2017, 173, 2743-2746.	0.7	23
871	Isolated congenital hepatic fibrosis associated with TMEM67 mutations: report of a new genotypeâ€“phenotype relationship. Clinical Case Reports (discontinued), 2017, 5, 1098-1102.	0.2	8
872	Lessons from the CAGIâ€4 Hopkins clinical panel challenge. Human Mutation, 2017, 38, 1155-1168.	1.1	6
873	Identification of Novel Breast Cancer Risk Loci. Cancer Research, 2017, 77, 5428-5437.	0.4	12
874	Early-Onset Alzheimer Disease and Candidate Risk Genes Involved in Endolysosomal Transport. JAMA Neurology, 2017, 74, 1113.	4.5	41
875	Establishing the role of rare coding variants in known Parkinson's disease risk loci. Neurobiology of Aging, 2017, 59, 220.e11-220.e18.	1.5	15

#	Article	IF	Citations
876	A commonSLC26A4-linked haplotype underlying non-syndromic hearing loss with enlargement of the vestibular aqueduct. Journal of Medical Genetics, 2017, 54, 665-673.	1.5	29
877	Constraints on eQTL Fine Mapping in the Presence of Multisite Local Regulation of Gene Expression. G3: Genes, Genomes, Genetics, 2017, 7, 2533-2544.	0.8	23
878	Rare, protein-truncating variants inATM,CHEK2andPALB2, but notXRCC2, are associated with increased breast cancer risks. Journal of Medical Genetics, 2017, 54, 732-741.	1.5	68
879	Aggregation of populationâ€based genetic variation over protein domain homologues and its potential use in genetic diagnostics. Human Mutation, 2017, 38, 1454-1463.	1.1	36
880	Whole-exome sequencing associates novel CSMD1 gene mutations with familial Parkinson disease. Neurology: Genetics, 2017, 3, e177.	0.9	27
881	MIRMMR: binary classification of microsatellite instability using methylation and mutations. Bioinformatics, 2017, 33, 3799-3801.	1.8	16
882	A syndromic extreme insulin resistance caused by biallelic POC1A mutations in exon 10. European Journal of Endocrinology, 2017, 177, K21-K27.	1.9	8
883	Blocking protein quality control to counter hereditary cancers. Genes Chromosomes and Cancer, 2017, 56, 823-831.	1.5	23
884	Settling the score: variant prioritization and Mendelian disease. Nature Reviews Genetics, 2017, 18, 599-612.	7.7	213
885	Neurodevelopmental Disorders Caused by De Novo Variants in KCNB1 Genotypes and Phenotypes. JAMA Neurology, 2017, 74, 1228.	4.5	79
886	Speech and Language: Translating the Genome. Trends in Genetics, 2017, 33, 642-656.	2.9	57
887	Efficient Prioritization of Multiple Causal eQTL Variants via Sparse Polygenic Modeling. Genetics, 2017, 207, 1301-1312.	1.2	10
888	Mitochondrial DNA Mutations Associated with Type 2 Diabetes Mellitus in Chinese Uyghur Population. Scientific Reports, 2017, 7, 16989.	1.6	29
889	Identification of MYLK3 mutations in familial dilated cardiomyopathy. Scientific Reports, 2017, 7, 17495.	1.6	34
890	The metabolic syndrome- associated small G protein ARL15 plays a role in adipocyte differentiation and adiponectin secretion. Scientific Reports, 2017, 7, 17593.	1.6	24
891	Resistance to Targeted Therapies in Breast Cancer. Resistance To Targeted Anti-cancer Therapeutics, 2017, , .	0.1	1
892	Identification of potentially oncogenic alterations from tumor-only samples reveals Fanconi anemia pathway mutations in bladder carcinomas. Npj Genomic Medicine, 2017, 2, 29.	1.7	14
893	Whole Exome Sequencing reveals new candidate genes in host genomic susceptibility to Respiratory Syncytial Virus Disease. Scientific Reports, 2017, 7, 15888.	1.6	29

#	Article	IF	Citations
894	Functional mapping and annotation of genetic associations with FUMA. Nature Communications, 2017, 8, 1826.	5.8	2,400
895	Functional germline variants as potential co-oncogenes. Npj Breast Cancer, 2017, 3, 46.	2.3	14
896	Novel KIAA0753 mutations extend the phenotype of skeletal ciliopathies. Scientific Reports, 2017, 7, 15585.	1.6	21
897	Genome-wide mapping of genetic determinants influencing DNA methylation and gene expression in human hippocampus. Nature Communications, 2017, 8, 1511.	5.8	60
898	Novel phenotypic variant in the MYH7 spectrum due to a stop-loss mutation in the C-terminal region: a case report. BMC Medical Genetics, 2017, 18, 105.	2.1	8
899	Multigeneration family with dominant SPG30 hereditary spastic paraplegia. Annals of Clinical and Translational Neurology, 2017, 4, 821-824.	1.7	19
900	Identification of deep intronic individual variants in patients with hemophilia A by nextâ€generation sequencing of the whole factor VIII gene. Research and Practice in Thrombosis and Haemostasis, 2017, 1, 264-274.	1.0	19
901	Germline variation in ADAMTSL1 is associated with prognosis following breast cancer treatment in young women. Nature Communications, 2017, 8, 1632.	5.8	18
902	Exome Pool-Seq in neurodevelopmental disorders. European Journal of Human Genetics, 2017, 25, 1364-1376.	1.4	77
903	Loss-of-activity-mutation in the cardiac chloride-bicarbonate exchanger AE3 causes short QT syndrome. Nature Communications, 2017, 8, 1696.	5.8	88
904	Clinical and Molecular Characterization of PMP22 point mutations in Taiwanese patients with Inherited Neuropathy. Scientific Reports, 2017, 7, 15363.	1.6	10
905	Percolation transition of cooperative mutational effects in colorectal tumorigenesis. Nature Communications, 2017, 8, 1270.	5.8	28
906	The contribution of rare variants to risk of schizophrenia in individuals with and without intellectual disability. Nature Genetics, 2017, 49, 1167-1173.	9.4	200
907	Investigating DNA-, RNA-, and protein-based features as a means to discriminate pathogenic synonymous variants. Human Mutation, 2017, 38, 1336-1347.	1.1	37
908	VEXOR: an integrative environment for prioritization of functional variants in fine-mapping analysis. Bioinformatics, 2017, 33, 1389-1391.	1.8	2
909	Integrative Genetic and Epigenetic Analysis Uncovers Regulatory Mechanisms of Autoimmune Disease. American Journal of Human Genetics, 2017, 101, 75-86.	2.6	29
910	Silencing of the Drosophila ortholog of SOX5 leads to abnormal neuronal development and behavioral impairment. Human Molecular Genetics, 2017, 26, 1472-1482.	1.4	42
911	A Massively Parallel Fluorescence Assay to Characterize the Effects of Synonymous Mutations on TP53 Expression. Molecular Cancer Research, 2017, 15, 1301-1307.	1.5	14

#	Article	IF	Citations
912	GTB â€“ an online genome tolerance browser. BMC Bioinformatics, 2017, 18, 20.	1.2	3
913	QueryOR: a comprehensive web platform for genetic variant analysis and prioritization. BMC Bioinformatics, 2017, 18, 225.	1.2	20
914	Comprehensive whole genome sequence analyses yields novel genetic and structural insights for Intellectual Disability. BMC Genomics, 2017, 18, 403.	1.2	15
915	A sequence-based method to predict the impact of regulatory variants using random forest. BMC Systems Biology, 2017, 11, 7.	3.0	13
916	CVE: an R package for interactive variant prioritisation in precision oncology. BMC Medical Genomics, 2017, 10, 37.	0.7	7
917	Not only dominant, not only optic atrophy: expanding the clinical spectrum associated with OPA1 mutations. Orphanet Journal of Rare Diseases, 2017, 12, 89.	1.2	39
918	Differentiated demographic histories and local adaptations between Sherpas and Tibetans. Genome Biology, 2017, 18, 115.	3.8	67
919	Lost in translation: returning germline genetic results in genome-scale cancer research. Genome Medicine, 2017, 9, 41.	3.6	27
920	Variant effect prediction tools assessed using independent, functional assay-based datasets: implications for discovery and diagnostics. Human Genomics, 2017, 11, 10.	1.4	68
921	A pipeline combining multiple strategies for prioritizing heterozygous variants for the identification of candidate genes in exome datasets. Human Genomics, 2017, 11, 11.	1.4	20
922	SEQSpark: A Complete Analysis Tool for Large-Scale Rare Variant Association Studies Using Whole-Genome and Exome Sequence Data. American Journal of Human Genetics, 2017, 101, 115-122.	2.6	9
923	Enrichment of low-frequency functional variants revealed by whole-genome sequencing of multiple isolated European populations. Nature Communications, 2017, 8, 15927.	5.8	64
924	Korean Variant Archive (KOVA): a reference database of genetic variations in the Korean population. Scientific Reports, 2017, 7, 4287.	1.6	60
925	PhD-SNPg: a webserver and lightweight tool for scoring single nucleotide variants. Nucleic Acids Research, 2017, 45, W247-W252.	6.5	132
926	Expanded repertoire of RASGRP2 variants responsible for platelet dysfunction and severe bleeding. Blood, 2017, 130, 1026-1030.	0.6	38
927	Impact of clinical exomes in neurodevelopmental and neurometabolic disorders. Molecular Genetics and Metabolism, 2017, 121, 297-307.	0.5	50
928	WDR73 missense mutation causes infantile onset intellectual disability and cerebellar hypoplasia in a consanguineous family. Clinica Chimica Acta, 2017, 464, 24-29.	0.5	10
929	A comprehensive strategy for exome-based preconception carrier screening. Genetics in Medicine, 2017, 19, 583-592.	1.1	30

#	Article	IF	Citations
930	A systematic comparison reveals substantial differences in chromosomal versus episomal encoding of enhancer activity. Genome Research, 2017, 27, 38-52.	2.4	244
931	NovelIRF6Mutations Detected in Orofacial Cleft Patients by Targeted Massively Parallel Sequencing. Journal of Dental Research, 2017, 96, 179-185.	2.5	12
932	HumanRECQHelicase Pathogenic Variants, Population Variation and â€œMissingâ€•Diseases. Human Mutation, 2017, 38, 193-203.	1.1	24
933	A Cell Type-Specific Expression Signature Predicts Haploinsufficient Autism-Susceptibility Genes. Human Mutation, 2017, 38, 204-215.	1.1	38
934	Multiple rare genetic variants coâ€segregating with familial IgA nephropathy all act within a single immuneâ€related network. Journal of Internal Medicine, 2017, 281, 189-205.	2.7	17
935	Ehlersâ€“Danlos syndrome with lethal cardiac valvular dystrophy in males carrying a novel splice mutation in FLNA. American Journal of Medical Genetics, Part A, 2017, 173, 169-176.	0.7	13
936	Lack of evidence for a role of genetic variation in TMEM230 in the risk for Parkinson's disease in the Caucasian population. Neurobiology of Aging, 2017, 50, 167.e11-167.e13.	1.5	24
937	In Silico Prediction of Deleteriousness for Nonsynonymous and Splice-Altering Single Nucleotide Variants in the Human Genome. Methods in Molecular Biology, 2017, 1498, 191-197.	0.4	17
938	A genotypic ascertainment approach to refute the association of MYO1A variants with non-syndromic deafness. European Journal of Human Genetics, 2017, 25, 147-149.	1.4	10
939	Acute Lung Injury and Repair. Respiratory Medicine, 2017, , .	0.1	1
940	Ciliopathy variant burden and developmental delay in children with hypoplastic left heart syndrome. Genetics in Medicine, 2017, 19, 711-714.	1.1	7
941	New era in genetics of early-onset muscle disease: Breakthroughs and challenges. Seminars in Cell and Developmental Biology, 2017, 64, 160-170.	2.3	24
942	Rare DNA variants in the brain-derived neurotrophic factor gene increase risk for attention-deficit hyperactivity disorder: a next-generation sequencing study. Molecular Psychiatry, 2017, 22, 580-584.	4.1	30
943	EFHC1 variants in juvenile myoclonic epilepsy: reanalysis according to NHGRI and ACMG guidelines for assigning disease causality. Genetics in Medicine, 2017, 19, 144-156.	1.1	34
944	Rare Genetic Variants of the Transthyretin Gene Are Associated with Alzheimerâ€™s Disease in Han Chinese. Molecular Neurobiology, 2017, 54, 5192-5200.	1.9	24
945	Whole exome sequencing identifies a homozygous nonsense variation in ALMS1 gene in a patient with syndromic obesity. Obesity Research and Clinical Practice, 2017, 11, 241-246.	0.8	14
946	mirVAFC: A Web Server for Prioritizations of Pathogenic Sequence Variants from Exome Sequencing Data via Classifications. Human Mutation, 2017, 38, 25-33.	1.1	3
947	Clinical exome sequencing in earlyâ€onset generalized dystonia and largeâ€scale resequencing followâ€up. Movement Disorders, 2017, 32, 549-559.	2.2	94

#	Article	IF	Citations
948	Atlas of human diseases influenced by genetic variants with extreme allele frequency differences. Human Genetics, 2017, 136, 39-54.	1.8	15
949	Clinical, imaging, pathological, and biochemical characterization of a novel presenilin 1 mutation (N135Y) causing Alzheimer's disease. Neurobiology of Aging, 2017, 49, 216.e7-216.e13.	1.5	22
950	Exome Sequencing Identified CCER2 as a Novel Candidate Gene for Moyamoya Disease. Journal of Stroke and Cerebrovascular Diseases, 2017, 26, 150-161.	0.7	13
951	A genome-wide association study identifies risk loci for childhood acute lymphoblastic leukemia at 10q26.13 and 12q23.1. Leukemia, 2017, 31, 573-579.	3.3	69
952	Expanding the genotypeâ€“phenotype spectrum in hereditary colorectal cancer by gene panel testing. Familial Cancer, 2017, 16, 195-203.	0.9	55
953	Targeted Nextâ€Generation Sequencing for Comprehensive Genetic Profiling of Pharmacogenes. Clinical Pharmacology and Therapeutics, 2017, 101, 396-405.	2.3	54
954	Vedolizumab as a successful treatment of CTLA-4â€“associated autoimmune enterocolitis. Journal of Allergy and Clinical Immunology, 2017, 139, 1043-1046.e5.	1.5	24
955	Genomic approaches to the assessment of human spina bifida risk. Birth Defects Research, 2017, 109, 120-128.	0.8	23
956	Whole exome sequencing identified sixty-five coding mutations in four neuroblastoma tumors. Scientific Reports, 2017, 7, 17787.	1.6	8
957	Excessive burden of lysosomal storage disorder gene variants in Parkinsonâ€™s disease. Brain, 2017, 140, 3191-3203.	3.7	323
958	A deep learning based scoring system for prioritizing susceptibility variants for mental disorders. , 2017, , .		2
959	Discovering the 3â€² UTR-mediated regulation of alpha-synuclein. Nucleic Acids Research, 2017, 45, 12888-12903.	6.5	32
960	Genetic variation in human drug-related genes. Genome Medicine, 2017, 9, 117.	3.6	104
961	Exploring functional variant using a deep learning framework. , 2017, , .		0
962	Exome-wide association study reveals novel psoriasis susceptibility locus at TNFSF15 and rare protective alleles in genes contributing to type I IFN signalling. Human Molecular Genetics, 2017, 26, 4301-4313.	1.4	41
963	Myelodysplasia and liver disease extend the spectrum of RTEL1 related telomeropathies. Haematologica, 2017, 102, e293-e296.	1.7	15
964	Deepâ€targeted sequencing of endothelial nitric oxide synthase gene exons uncovers exercise intensity and ethnicityâ€dependent associations with postâ€exercise hypotension. Physiological Reports, 2017, 5, e13510.	0.7	9
965	Rare GABRA3 variants are associated with epileptic seizures, encephalopathy and dysmorphic features. Brain, 2017, 140, 2879-2894.	3.7	33

#	Article	IF	Citations
966	A molecular inversion probe-based next-generation sequencing panel to detect germline mutations in Chinese early-onset colorectal cancer patients. Oncotarget, 2017, 8, 24533-24547.	0.8	12
968	Warfarin Anticoagulation Therapy in Caribbean Hispanics of Puerto Rico: A Candidate Gene Association Study. Frontiers in Pharmacology, 2017, 8, 347.	1.6	18
969	Candidate Gene Resequencing in a Large Bicuspid Aortic Valve-Associated Thoracic Aortic Aneurysm Cohort: SMAD6 as an Important Contributor. Frontiers in Physiology, 2017, 8, 400.	1.3	85
970	When loss-of-function is loss of function: assessing mutational signatures and impact of loss-of-function genetic variants. Bioinformatics, 2017, 33, i389-i398.	1.8	53
971	Identification of candidate protective variants for common diseases and evaluation of their protective potential. BMC Genomics, 2017, 18, 575.	1.2	15
972	Development of pathogenicity predictors specific for variants that do not comply with clinical guidelines for the use of computational evidence. BMC Genomics, 2017, 18, 569.	1.2	14
973	Associating mutations causing cystinuria with disease severity with the aim of providing precision medicine. BMC Genomics, 2017, 18, 550.	1.2	16
974	Accurate and reproducible functional maps in 127 human cell types via 2D genome segmentation. Nucleic Acids Research, 2017, 45, 9823-9836.	6.5	42
975	Population-Specific Associations of Deleterious Rare Variants in Coding Region of P2RY1â€“P2RY12 Purinergic Receptor Genes in Large-Vessel Ischemic Stroke Patients. International Journal of Molecular Sciences, 2017, 18, 2678.	1.8	10
976	Diagnostic Yield of Whole Exome Sequencing in Pediatric Dilated Cardiomyopathy. Journal of Cardiovascular Development and Disease, 2017, 4, 11.	0.8	37
977	Independent impacts of aging on mitochondrial DNA quantity and quality in humans. BMC Genomics, 2017, 18, 890.	1.2	116
978	Whole Genome Sequencing Revealed Mutations in Two Independent Genes as the Underlying Cause of Retinal Degeneration in an Ashkenazi Jewish Pedigree. Genes, 2017, 8, 210.	1.0	14
979	An Expanded Multi-Organ Disease Phenotype Associated with Mutations in YARS. Genes, 2017, 8, 381.	1.0	19
980	Big Data Analytics for Genomic Medicine. International Journal of Molecular Sciences, 2017, 18, 412.	1.8	121
981	Molecular Autopsy for Sudden Death in the Young: Is Data Aggregation the Key?. Frontiers in Cardiovascular Medicine, 2017, 4, 72.	1.1	26
982	Epigenetic Combinatorial Patterns Predict Disease Variants. Frontiers in Genetics, 2017, 8, 71.	1.1	2
983	A Review of Pathway-Based Analysis Tools That Visualize Genetic Variants. Frontiers in Genetics, 2017, 8, 174.	1.1	67
984	Clinical and Molecular Heterogeneity of RTEL1 Deficiency. Frontiers in Immunology, 2017, 8, 449.	2.2	35

#	Article	IF	Citations
985	Novel Combined Immune Deficiency and Radiation Sensitivity Blended Phenotype in an Adult with Biallelic Variations in ZAP70 and RNF168. Frontiers in Immunology, 2017, 8, 576.	2.2	23
986	Targeted Genetic Screen in Amyotrophic Lateral Sclerosis Reveals Novel Genetic Variants with Synergistic Effect on Clinical Phenotype. Frontiers in Molecular Neuroscience, 2017, 10, 370.	1.4	24
987	Resources for Interpreting Variants in Precision Genomic Oncology Applications. Frontiers in Oncology, 2017, 7, 214.	1.3	18
988	Up, Down, and All Around: Diagnosis and Treatment of Novel STAT3 Variant. Frontiers in Pediatrics, 2017, 5, 49.	0.9	25
989	The Genomics of Neonatal Abstinence Syndrome. Frontiers in Pediatrics, 2017, 5, 176.	0.9	13
990	Idiopathic hypereosinophilia is clonal disorder? Clonality identified by targeted sequencing. PLoS ONE, 2017, 12, e0185602.	1.1	27
991	Targeted high throughput sequencing in hereditary ataxia and spastic paraplegia. PLoS ONE, 2017, 12, e0174667.	1.1	54
992	Alport syndrome cold cases: Missing mutations identified by exome sequencing and functional analysis. PLoS ONE, 2017, 12, e0178630.	1.1	16
993	Whole exome sequencing in thrombophilic pedigrees to identify genetic risk factors for venous thromboembolism. PLoS ONE, 2017, 12, e0187699.	1.1	14
994	New NR5A1 mutations and phenotypic variations of gonadal dysgenesis. PLoS ONE, 2017, 12, e0176720.	1.1	37
995	Functional significance of rare neuroligin 1 variants found in autism. PLoS Genetics, 2017, 13, e1006940.	1.5	76
996	Semantic prioritization of novel causative genomic variants. PLoS Computational Biology, 2017, 13, e1005500.	1.5	28
997	Fetal de novo mutations and preterm birth. PLoS Genetics, 2017, 13, e1006689.	1.5	30
998	Human mutations in integrator complex subunits link transcriptome integrity to brain development. PLoS Genetics, 2017, 13, e1006809.	1.5	66
999	Rare coding variants pinpoint genes that control human hematological traits. PLoS Genetics, 2017, 13, e1006925.	1.5	39
1000	Systematic tissue-specific functional annotation of the human genome highlights immune-related DNA elements for late-onset Alzheimerâ€™s disease. PLoS Genetics, 2017, 13, e1006933.	1.5	96
1001	Incomplete dominance of deleterious alleles contributes substantially to trait variation and heterosis in maize. PLoS Genetics, 2017, 13, e1007019.	1.5	136
1002	Integrated rare variant-based risk gene prioritization in disease case-control sequencing studies. PLoS Genetics, 2017, 13, e1007142.	1.5	7

#	Article	IF	Citations
1003	Whole exome sequencing in the Framingham Heart Study identifies rare variation in HYAL2 that influences platelet aggregation. Thrombosis and Haemostasis, 2017, 117, 1083-1092.	1.8	11
1004	Whole exome sequencing in 342 congenital cardiac left sided lesion cases reveals extensive genetic heterogeneity and complex inheritance patterns. Genome Medicine, 2017, 9, 95.	3.6	37
1005	Using the Neandertal genome to study the evolution of small insertions and deletions in modern humans. BMC Evolutionary Biology, 2017, 17, 179.	3.2	14
1006	Spectrum of mutations in monogenic diabetes genes identified from high-throughput DNA sequencing of 6888 individuals. BMC Medicine, 2017, 15, 213.	2.3	75
1007	Genome-wide gene by lead exposure interaction analysis identifies UNC5D as a candidate gene for neurodevelopment. Environmental Health, 2017, 16, 81.	1.7	20
1008	Exome sequencing characterizes the somatic mutation spectrum of early serrated lesions in a patient with serrated polyposis syndrome (SPS). Hereditary Cancer in Clinical Practice, 2017, 15, 22.	0.6	6
1009	Recurrent de novo mutations in neurodevelopmental disorders: properties and clinical implications. Genome Medicine, 2017, 9, 101.	3.6	112
1010	Using DIVAN to assess disease/trait-associated single nucleotide variants in genome-wide scale. BMC Research Notes, 2017, 10, 530.	0.6	10
1011	A patient with van Maldergem syndrome with endocrine abnormalities, hypogonadotropic hypogonadism, and breast aplasia/hypoplasia. International Journal of Pediatric Endocrinology (Springer), 2017, 2017, 12.	1.6	4
1012	Evaluation of in silico algorithms for use with ACMG/AMP clinical variant interpretation guidelines. Genome Biology, 2017, 18, 225.	3.8	185
1013	Rare non-coding variants are associated with plasma lipid traits in a founder population. Scientific Reports, 2017, 7, 16415.	1.6	31
1014	Variant Ranker: a web-tool to rank genomic data according to functional significance. BMC Bioinformatics, 2017, 18, 341.	1.2	21
1015	An integrative approach to predicting the functional effects of small indels in non-coding regions of the human genome. BMC Bioinformatics, 2017, 18, 442.	1.2	34
1016	Predicting enhancers with deep convolutional neural networks. BMC Bioinformatics, 2017, 18, 478.	1.2	76
1017	PON-SC â€“ program for identifying steric clashes caused by amino acid substitutions. BMC Bioinformatics, 2017, 18, 531.	1.2	7
1018	Canary: an atomic pipeline for clinical amplicon assays. BMC Bioinformatics, 2017, 18, 555.	1.2	4
1019	A practical guide to filtering and prioritizing genetic variants. BioTechniques, 2017, 62, 18-30.	0.8	57
1020	Exome Sequencing of Extended Families with Alzheimerâ€™s Disease Identifies Novel Genes Implicated in Cell Immunity and Neuronal Function. , 2017, 07, .		13

#	Article	IF	Citations
1021	Utility of Population-Level DNA Sequence Data in the Diagnosis of Hereditary Endocrine Disease. Journal of the Endocrine Society, 2017, 1, 1507-1526.	0.1	15
1022	CERENKOV. , 2017, , .		3
1023	Effects of Type 1 Diabetes Risk Alleles on Immune Cell Gene Expression. Genes, 2017, 8, 167.	1.0	17
1024	Novel Complex ABCA4 Alleles in Brazilian Patients With Stargardt Disease: Genotypeâ€“Phenotype Correlation. , 2017, 58, 5723.		12
1026	Identifying noncoding risk variants using disease-relevant gene regulatory networks. Nature Communications, 2018, 9, 702.	5.8	35
1027	Exome sequencing reveals blended phenotype of double heterozygous FBN1 and FBN2 variants in a fetus. European Journal of Medical Genetics, 2018, 61, 399-402.	0.7	8
1028	Single suture craniosynostosis: Identification of rare variants in genes associated with syndromic forms. American Journal of Medical Genetics, Part A, 2018, 176, 290-300.	0.7	34
1029	Applications of neural networks to the studies of phase transitions of two-dimensional Potts models. Annals of Physics, 2018, 391, 312-331.	1.0	43
1030	Inborn Errors of RNA Lariat Metabolism in Humans with Brainstem Viral Infection. Cell, 2018, 172, 952-965.e18.	13.5	92
1031	Somatic mutagenesis in satellite cells associates with human skeletal muscle aging. Nature Communications, 2018, 9, 800.	5.8	94
1032	Network analysis of coronary artery disease risk genes elucidates disease mechanisms and druggable targets. Scientific Reports, 2018, 8, 3434.	1.6	43
1033	Genetics of dementia in a Finnish cohort. European Journal of Human Genetics, 2018, 26, 827-837.	1.4	6
1034	The human noncoding genome defined by genetic diversity. Nature Genetics, 2018, 50, 333-337.	9.4	137
1035	Whole-Genome Sequencing of Pharmacogenetic Drug Response in Racially Diverse Children with Asthma. American Journal of Respiratory and Critical Care Medicine, 2018, 197, 1552-1564.	2.5	102
1036	Recessive mutations in VPS13D cause childhood onset movement disorders. Annals of Neurology, 2018, 83, 1089-1095.	2.8	104
1037	Kidney enlargement and multiple liver cyst formation implicate mutations in PKD1/2 in adult sporadic polycystic kidney disease. Clinical Genetics, 2018, 94, 125-131.	1.0	16
1038	Wholeâ€exome sequencing improves genetic testing accuracy in pulmonary artery hypertension. Pulmonary Circulation, 2018, 8, 1-9.	0.8	3
1039	Genetic Variation in Genes Underlying Diverse Dementias May Explain a Small Proportion of Cases in the Alzheimerâ€™s Disease Sequencing Project. Dementia and Geriatric Cognitive Disorders, 2018, 45, 1-17.	0.7	22

#	Article	IF	Citations
1040	Genotypic effect of a mutation of the <scp>MYBPC</scp>3 gene and two phenotypes with different patterns of inheritance. Journal of Clinical Laboratory Analysis, 2018, 32, e22419.	0.9	6
1041	Analysis of DICER1 in familial and sporadic cases of transposition of the great arteries. Congenital Heart Disease, 2018, 13, 401-406.	0.0	2
1042	Genotype, phenotype and in silico pathogenicity analysis of HEXB mutations: Panel based sequencing for differential diagnosis of gangliosidosis. Clinical Neurology and Neurosurgery, 2018, 167, 43-53.	0.6	4
1043	Similarity Network Fusion. Rheumatic Disease Clinics of North America, 2018, 44, 285-293.	0.8	5
1044	Rare variants in axonogenesis genes connect three families with soundâ€“color synesthesia. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 3168-3173.	3.3	34
1045	GNAO1 Mutationâ€“Induced Pediatric Dystonic Storm Rescue With Pallidal Deep Brain Stimulation. Journal of Child Neurology, 2018, 33, 413-416.	0.7	31
1046	De novo hotspot variants in CYFIP2 cause earlyâ€onset epileptic encephalopathy. Annals of Neurology, 2018, 83, 794-806.	2.8	60
1047	No novel, high penetrant gene might remain to be found in Japanese patients with unknown MODY. Journal of Human Genetics, 2018, 63, 821-829.	1.1	3
1048	Further audiovestibular characterization of DFNB77, caused by deleterious variants in LOXHD1, and investigation into the involvement of Fuchs corneal dystrophy. Clinical Genetics, 2018, 94, 221-231.	1.0	11
1049	Functional relevance of synonymous alleles reflected in allele rareness in the population. Genomics, 2018, 110, 347-354.	1.3	4
1050	Autosomal recessive otofaciocervical syndrome type 2 with novel homozygous small insertion in PAX1 gene. American Journal of Medical Genetics, Part A, 2018, 176, 1200-1206.	0.7	21
1051	Genome sequencing reveals a novel genetic mechanism underlying dihydropyrimidine dehydrogenase deficiency: A novel missense variant c.1700G>A and a large intragenic inversion in DPYD spanning intron 8 to intron 12. Human Mutation, 2018, 39, 947-953.	1.1	6
1052	Biological Insights Into Muscular Strength: Genetic Findings in the UK Biobank. Scientific Reports, 2018, 8, 6451.	1.6	78
1053	Whole genome sequencing of Caribbean Hispanic families with lateâ€onset Alzheimer's disease. Annals of Clinical and Translational Neurology, 2018, 5, 406-417.	1.7	42
1054	Systematic reanalysis of genomic data improves quality of variant interpretation. Clinical Genetics, 2018, 94, 174-178.	1.0	30
1055	Genomic analysis identifies masqueraders of fullâ€term cerebral palsy. Annals of Clinical and Translational Neurology, 2018, 5, 538-551.	1.7	73
1056	Genetic variations and risk of placental abruption: A genome-wide association study and meta-analysis of genome-wide association studies. Placenta, 2018, 66, 8-16.	0.7	15
1057	Point-of-care whole-exome sequencing of idiopathic male infertility. Genetics in Medicine, 2018, 20, 1365-1373.	1.1	105

#	Article	IF	Citations
1058	Structural dynamics is a determinant of the functional significance of missense variants. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 4164-4169.	3.3	76
1059	De novo mutations of the ATP6V1A gene cause developmental encephalopathy with epilepsy. Brain, 2018, 141, 1703-1718.	3.7	69
1060	TiSAn: estimating tissue-specific effects of coding and non-coding variants. Bioinformatics, 2018, 34, 3061-3068.	1.8	5
1061	Germline Genetic IKZF1 Variation and Predisposition to Childhood Acute Lymphoblastic Leukemia. Cancer Cell, 2018, 33, 937-948.e8.	7.7	142
1062	Physiological and Genetic Adaptations to Diving in Sea Nomads. Cell, 2018, 173, 569-580.e15.	13.5	129
1063	Opportunities and obstacles for deep learning in biology and medicine. Journal of the Royal Society Interface, 2018, 15, 20170387.	1.5	1,282
1064	Comprehensive Characterization of Cancer Driver Genes and Mutations. Cell, 2018, 173, 371-385.e18.	13.5	1,670
1065	Genetic variants in mRNA untranslated regions. Wiley Interdisciplinary Reviews RNA, 2018, 9, e1474.	3.2	118
1066	Identification of rare heterozygous missense mutations in FANCA in esophageal atresia patients using next-generation sequencing. Gene, 2018, 661, 182-188.	1.0	2
1067	VarCards: an integrated genetic and clinical database for coding variants in the human genome. Nucleic Acids Research, 2018, 46, D1039-D1048.	6.5	148
1068	Targeted Next-generation Sequencing and Bioinformatics Pipeline to Evaluate Genetic Determinants of Constitutional Disease. Journal of Visualized Experiments, 2018, , .	0.2	17
1069	Pleiotropic mapping and annotation selection in genome-wide association studies with penalized Gaussian mixture models. Bioinformatics, 2018, 34, 2797-2807.	1.8	34
1070	Compound heterozygosity for loss-of-function FARSB variants in a patient with classic features of recessive aminoacyl-tRNA synthetase-related disease. Human Mutation, 2018, 39, 834-840.	1.1	30
1071	CommonVWFsequence variants associated with higher VWF and FVIII are less frequent in subjects diagnosed with type 1 VWD. Research and Practice in Thrombosis and Haemostasis, 2018, 2, 390-398.	1.0	5
1072	Clinical and biochemical features of different molecular etiologies of familial chylomicronemia. Journal of Clinical Lipidology, 2018, 12, 920-927.e4.	0.6	97
1073	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
1074	Genetic and mechanistic diversity in pediatric hemophagocytic lymphohistiocytosis. Blood, 2018, 132, 89-100.	0.6	139
1075	Inference of the human polyadenylation code. Bioinformatics, 2018, 34, 2889-2898.	1.8	26

#	Article	IF	Citations
1076	Deep learning of genomic variation and regulatory network data. Human Molecular Genetics, 2018, 27, R63-R71.	1.4	64
1077	Exome Sequencing in Children With Pulmonary Arterial Hypertension Demonstrates Differences Compared With Adults. Circulation Genomic and Precision Medicine, 2018, 11, e001887.	1.6	104
1078	Inferring the effect of genomic variation in the new era of genomics. Human Mutation, 2018, 39, 756-773.	1.1	24
1079	Genotypeâ€Phenotype Relations for the Parkinson's Disease Genes Parkin, PINK1, DJ1: MDSGene Systematic Review. Movement Disorders, 2018, 33, 730-741.	2.2	215
1080	COQ2 variants in Parkinsonâ€™s disease and multiple system atrophy. Journal of Neural Transmission, 2018, 125, 937-944.	1.4	10
1081	Identification of rare sequence variation underlying heritable pulmonary arterial hypertension. Nature Communications, 2018, 9, 1416.	5.8	279
1082	Molecular analysis of PALB2â€associated breast cancers. Journal of Pathology, 2018, 245, 53-60.	2.1	46
1083	A phenotype centric benchmark of variant prioritisation tools. Npj Genomic Medicine, 2018, 3, 5.	1.7	39
1084	Identification of a novel homozygous UNC80 variant in a child with infantile hypotonia with psychomotor retardation and characteristic facies-2 (IHPRF2). Metabolic Brain Disease, 2018, 33, 869-873.	1.4	11
1085	Defective RNA sensing by RIG-I in severe influenza virus infection. Clinical and Experimental Immunology, 2018, 192, 366-376.	1.1	39
1086	Yield of the RYR2 Genetic Test in Suspected Catecholaminergic Polymorphic Ventricular Tachycardia and Implications for Test Interpretation. Circulation Genomic and Precision Medicine, 2018, 11, e001424.	1.6	30
1087	Functional Dysregulation of CDC42 Causes Diverse Developmental Phenotypes. American Journal of Human Genetics, 2018, 102, 309-320.	2.6	138
1088	The first two confirmed sub-Saharan African families with germline TP53 mutations causing Li-Fraumeni syndrome. Familial Cancer, 2018, 17, 607-613.	0.9	6
1089	Bioinformatics analysis of non-synonymous variants in the KLF genes related to cardiac diseases. Gene, 2018, 650, 68-76.	1.0	5
1090	Pathogenic significance of SCN1A splicing variants causing Dravet syndrome: Improving diagnosis with targeted sequencing for variants by in silico analysis. Clinical Neurology and Neurosurgery, 2018, 166, 80-90.	0.6	2
1091	Prediction of factor VIII inhibitor development in the SIPPET cohort by mutational analysis and factor VIII antigen measurement. Journal of Thrombosis and Haemostasis, 2018, 16, 778-790.	1.9	23
1092	GWAS-based machine learning approach to predict duloxetine response in major depressive disorder. Journal of Psychiatric Research, 2018, 99, 62-68.	1.5	60
1093	Somatic APC mosaicism and oligogenic inheritance in genetically unsolved colorectal adenomatous polyposis patients. European Journal of Human Genetics, 2018, 26, 387-395.	1.4	26

#	Article	IF	Citations
1094	Cancer genetics meets biomolecular mechanismâ€”bridging an ageâ€old gulf. FEBS Letters, 2018, 592, 463-474.	1.3	9
1095	Combined annotationâ€dependent depletion score for BRCA1/2 variants in patients with breast and/or ovarian cancer. Cancer Science, 2018, 109, 453-461.	1.7	17
1096	Disclosure of cardiac variants of uncertain significance results in an exome cohort. Clinical Genetics, 2018, 93, 1022-1029.	1.0	17
1097	Novel and recurrent RNF213 variants in Japanese pediatric patients with moyamoya disease. Human Genome Variation, 2018, 5, 17060.	0.4	12
1098	Genetic basis of cardiomyopathy and the genotypes involved in prognosis and left ventricular reverse remodeling. Scientific Reports, 2018, 8, 1998.	1.6	94
1099	IW-Scoring: an Integrative Weighted Scoring framework for annotating and prioritizing genetic variations in the noncoding genome. Nucleic Acids Research, 2018, 46, e47-e47.	6.5	30
1100	Rare Variants in Tissue Inhibitor of Metalloproteinase 2 as a Risk Factor for Schizophrenia: Evidence From Familial and Cohort Analysis. Schizophrenia Bulletin, 2019, 45, 256-263.	2.3	12
1101	A robust targeted sequencing approach for low input and variable quality DNA from clinical samples. Npj Genomic Medicine, 2018, 3, 2.	1.7	20
1102	Robust identification of mosaic variants in congenital heart disease. Human Genetics, 2018, 137, 183-193.	1.8	43
1103	Diagnostic utility of exome sequencing in the evaluation of neuromuscular disorders. Neurology: Genetics, 2018, 4, e212.	0.9	42
1104	The emerging genetic landscape of cerebral palsy. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 147, 331-342.	1.0	23
1105	Orchid: a novel management, annotation and machine learning framework for analyzing cancer mutations. Bioinformatics, 2018, 34, 936-942.	1.8	16
1106	High Yield of Pathogenic Germline Mutations Causative or Likely Causative of the Cancer Phenotype in Selected Children with Cancer. Clinical Cancer Research, 2018, 24, 1594-1603.	3.2	52
1107	Exome-wide analysis of mutational burden in patients with typical and atypical Rolandic epilepsy. European Journal of Human Genetics, 2018, 26, 258-264.	1.4	22
1108	Re-analysis of public genetic data reveals a rare X-chromosomal variant associated with type 2 diabetes. Nature Communications, 2018, 9, 321.	5.8	85
1109	Inherited Cardiovascular Conditions: Phenotype-Genotype Data Mining and Sharing, and Databases. , 2018, , 869-877.		1
1110	De novo mutations in GRIN1 cause extensive bilateral polymicrogyria. Brain, 2018, 141, 698-712.	3.7	72
1111	Further Investigation of the Role of ACYP2 and WFS1 Pharmacogenomic Variants in the Development of Cisplatin-Induced Ototoxicity in Testicular Cancer Patients. Clinical Cancer Research, 2018, 24, 1866-1871.	3.2	32

#	Article	IF	Citations
1112	Constitutional SAMD9L mutations cause familial myelodysplastic syndrome and transient monosomy 7. Haematologica, 2018, 103, 427-437.	1.7	83
1113	Neurodegeneration with brain iron accumulation 2A: Report of four independent cases. Meta Gene, 2018, 15, 87-89.	0.3	0
1114	NOX1 loss-of-function genetic variants in patients with inflammatory bowel disease. Mucosal Immunology, 2018, 11, 562-574.	2.7	71
1115	A child with Myhre syndrome presenting with corectopia and tetralogy of Fallot. American Journal of Medical Genetics, Part A, 2018, 176, 426-430.	0.7	15
1116	â€œLowe syndrome: A particularly severe phenotype without clinical kidney involvementâ€• American Journal of Medical Genetics, Part A, 2018, 176, 460-464.	0.7	4
1117	Quantitative Missense Variant Effect Prediction Using Large-Scale Mutagenesis Data. Cell Systems, 2018, 6, 116-124.e3.	2.9	176
1118	Frequency and characteristics of the TBK1 gene variants in Japanese patients with sporadic amyotrophic lateral sclerosis. Neurobiology of Aging, 2018, 64, 158.e15-158.e19.	1.5	15
1119	Disease-causing mutations in the promoter and enhancer of the ornithine transcarbamylase gene. Human Mutation, 2018, 39, 527-536.	1.1	37
1120	Principles and methods of in-silico prioritization of non-coding regulatory variants. Human Genetics, 2018, 137, 15-30.	1.8	37
1121	Tools for protein science. Protein Science, 2018, 27, 6-9.	3.1	2
1122	MMAB, a novel candidate gene to be screened in the molecular diagnosis of Mevalonate Kinase Deficiency. Rheumatology International, 2018, 38, 121-127.	1.5	1
1123	Mutations in SELENBP1, encoding a novel human methanethiol oxidase, cause extraoral halitosis. Nature Genetics, 2018, 50, 120-129.	9.4	86
1124	Human genetics of infectious diseases: Unique insights into immunological redundancy. Seminars in Immunology, 2018, 36, 1-12.	2.7	82
1125	Proteinâ€altering variants of <scp>PTPN</scp>2 in childhoodâ€onset Type 1A diabetes. Diabetic Medicine, 2018, 35, 376-380.	1.2	7
1126	Infants with Atypical Presentations of Alveolar Capillary Dysplasia with Misalignment of the Pulmonary Veins Who Underwent Bilateral Lung Transplantation. Journal of Pediatrics, 2018, 194, 158-164.e1.	0.9	48
1127	Putative functional genes in idiopathic dilated cardiomyopathy. Scientific Reports, 2018, 8, 66.	1.6	7
1128	A benchmark study of scoring methods for non-coding mutations. Bioinformatics, 2018, 34, 1635-1641.	1.8	21
1129	Rare loss of function mutations in N-methyl-d-aspartate glutamate receptors and their contributions to schizophrenia susceptibility. Translational Psychiatry, 2018, 8, 12.	2.4	41

#	Article	IF	Citations
1130	Arterial tortuosity syndrome: 40 new families and literature review. Genetics in Medicine, 2018, 20, 1236-1245.	1.1	66
1131	Variation in Cilia Protein Genes and Progression of Lung Disease in Cystic Fibrosis. Annals of the American Thoracic Society, 2018, 15, 440-448.	1.5	14
1132	Exome-wide rare variant analyses of two bone mineral density phenotypes: the challenges of analyzing rare genetic variation. Scientific Reports, 2018, 8, 220.	1.6	2
1133	De novo variants in SETD1B are associated with intellectual disability, epilepsy and autism. Human Genetics, 2018, 137, 95-104.	1.8	60
1134	Modeling mutant/wild-type interactions to ascertain pathogenicity of PROKR2 missense variants in patients with isolated GnRH deficiency. Human Molecular Genetics, 2018, 27, 338-350.	1.4	21
1135	Nextâ€generation metrics for monitoring genetic erosion within populations of conservation concern. Evolutionary Applications, 2018, 11, 1066-1083.	1.5	93
1136	SNPDelScore: combining multiple methods to score deleterious effects of noncoding mutations in the human genome. Bioinformatics, 2018, 34, 289-291.	1.8	11
1137	Improvement of Self-Injury With Dopamine and Serotonin Replacement Therapy in a Patient With a Hemizygous PAK3 Mutation: A New Therapeutic Strategy for Neuropsychiatric Features of an Intellectual Disability Syndrome. Journal of Child Neurology, 2018, 33, 106-113.	0.7	20
1138	De novo mutations implicate novel genes in systemic lupus erythematosus. Human Molecular Genetics, 2018, 27, 421-429.	1.4	52
1139	DNAAF1 links heart laterality with the AAA+ ATPase RUVBL1 and ciliary intraflagellar transport. Human Molecular Genetics, 2018, 27, 529-545.	1.4	45
1140	Loss-of-function mutations inISCA2disrupt 4Fe-4S cluster machinery and cause a fatal leukodystrophy with hyperglycinemia and mtDNA depletion. Human Mutation, 2018, 39, 537-549.	1.1	21
1141	Exome Sequencing and Genotyping Identify a Rare Variant inNLRP7Gene Associated With Ulcerative Colitis. Journal of Crohn's and Colitis, 2018, 12, 321-326.	0.6	14
1142	Human pain genetics database: a resource dedicated to human pain genetics research. Pain, 2018, 159, 749-763.	2.0	80
1143	Genetic analysis of CHARGE syndrome identifies overlapping molecular biology. Genetics in Medicine, 2018, 20, 1022-1029.	1.1	43
1144	FUN-LDA: A Latent Dirichlet Allocation Model for Predicting Tissue-Specific Functional Effects of Noncoding Variation: Methods and Applications. American Journal of Human Genetics, 2018, 102, 920-942.	2.6	75
1145	Promoter of lncRNA Gene PVT1 Is a Tumor-Suppressor DNA Boundary Element. Cell, 2018, 173, 1398-1412.e22.	13.5	362
1146	Genetic screening of male patients with primary hypogammaglobulinemia can guide diagnosis and clinical management. Human Immunology, 2018, 79, 571-577.	1.2	5
1147	Cross-linking BioThings APIs through JSON-LD to facilitate knowledge exploration. BMC Bioinformatics, 2018, 19, 30.	1.2	24

#	Article	IF	Citations
1148	Whole exome sequencing in three families segregating a pediatric case of sarcoidosis. BMC Medical Genomics, 2018, 11, 23.	0.7	26
1149	Complex genetic architecture in severe hypobetalipoproteinemia. Lipids in Health and Disease, 2018, 17, 48.	1.2	14
1150	Evaluating the breast cancer predisposition role of rare variants in genes associated with low-penetrance breast cancer risk SNPs. Breast Cancer Research, 2018, 20, 3.	2.2	19
1151	The use of whole exome sequencing and murine patient derived xenografts as a method of chemosensitivity testing in sarcoma. Clinical Sarcoma Research, 2018, 8, 4.	2.3	4
1152	A Mixed-Effects Model for Powerful Association Tests in Integrative Functional Genomics. American Journal of Human Genetics, 2018, 102, 904-919.	2.6	30
1153	Phenotypes in siblings with homozygous mutations of <scp>TRAPPC</scp>9 and/or <scp>MCPH</scp>1 support a bifunctional model of <scp>MCPH</scp>1. Molecular Genetics & Genomic Medicine, 2018, 6, 660-665.	0.6	15
1154	GenomicScores: seamless access to genomewide position-specific scores from R and Bioconductor. Bioinformatics, 2018, 34, 3208-3210.	1.8	22
1155	MoBiDiC Prioritization Algorithm, a Free, Accessible, and Efficient Pipeline for Single-Nucleotide Variant Annotation and Prioritization for Next-Generation Sequencing Routine Molecular Diagnosis. Journal of Molecular Diagnostics, 2018, 20, 465-473.	1.2	13
1156	Functional missense and splicing variants in the retinoic acid catabolizing enzyme CYP26C1 in idiopathic short stature. European Journal of Human Genetics, 2018, 26, 1113-1120.	1.4	10
1157	Patterns of Genetic Coding Variation in a Native American Population before and after European Contact. American Journal of Human Genetics, 2018, 102, 806-815.	2.6	33
1158	Whole exome sequencing in an Italian family with isolated maxillary canine agenesis and canine eruption anomalies. Archives of Oral Biology, 2018, 91, 96-102.	0.8	8
1159	Whole-Exome Sequencing in Age-Related Macular Degeneration Identifies Rare Variants in COL8A1, a Component of Bruchâ€™s Membrane. Ophthalmology, 2018, 125, 1433-1443.	2.5	35
1160	DeFine: deep convolutional neural networks accurately quantify intensities of transcription factor-DNA binding and facilitate evaluation of functional non-coding variants. Nucleic Acids Research, 2018, 46, e69-e69.	6.5	89
1161	A Homozygous LAMA2 Mutation of c.818G>A Caused Partial Merosin Deficiency in a Japanese Patient. Internal Medicine, 2018, 57, 877-882.	0.3	6
1162	Mutations in SZT2 result in earlyâ€onset epileptic encephalopathy and leukoencephalopathy. American Journal of Medical Genetics, Part A, 2018, 176, 1443-1448.	0.7	15
1163	Keratoendotheliitis Fugax Hereditaria: A Novel Cryopyrin-Associated Periodic Syndrome Caused by a Mutation in the Nucleotide-Binding Domain, Leucine-Rich Repeat Family, Pyrin Domain-Containing 3 () Tj ETQq1 1 0.784314 rgBT /Ov	1.7	
1164	Genome Sequencing and RNA-Motif Analysis Reveal Novel Damaging Noncoding Mutations in Human Tumors. Molecular Cancer Research, 2018, 16, 1112-1124.	1.5	18
1165	A new mutation in steroidogenic acute regulatory protein (StAR) is segregated in an Iranian family. Meta Gene, 2018, 16, 196-198.	0.3	2

#	Article	IF	Citations
1166	New workflow for classification of genetic variantsâ€™ pathogenicity applied to hereditary recurrent fevers by the International Study Group for Systemic Autoinflammatory Diseases (INSAID). Journal of Medical Genetics, 2018, 55, 530-537.	1.5	117
1167	Germline Variants in the POT1-Gene in High-Risk Melanoma Patients in Austria. G3: Genes, Genomes, Genetics, 2018, 8, 1475-1480.	0.8	12
1168	Relationship between Deleterious Variation, Genomic Autozygosity, and Disease Risk: Insights from The 1000 Genomes Project. American Journal of Human Genetics, 2018, 102, 658-675.	2.6	29
1169	Targeted genetic analysis in a large cohort of familial and sporadic cases of aneurysm or dissection of the thoracic aorta. Genetics in Medicine, 2018, 20, 1414-1422.	1.1	48
1170	Phenotype risk scores identify patients with unrecognized Mendelian disease patterns. Science, 2018, 359, 1233-1239.	6.0	164
1171	Hypertrophy Regression With N-Acetylcysteine in Hypertrophic Cardiomyopathy (HALT-HCM). Circulation Research, 2018, 122, 1109-1118.	2.0	42
1172	Functional annotation of genomic variants in studies of late-onset Alzheimerâ€™s disease. Bioinformatics, 2018, 34, 2724-2731.	1.8	30
1173	Intestinal Failure and Aberrant Lipid Metabolism in Patients WithÂDGAT1 Deficiency. Gastroenterology, 2018, 155, 130-143.e15.	0.6	83
1174	Whole-exome sequencing reanalysis at 12 months boosts diagnosis and is cost-effective when applied early in Mendelian disorders. Genetics in Medicine, 2018, 20, 1564-1574.	1.1	132
1175	A Screening Approach to Identify Clinically Actionable Variants Causing Congenital Heart Disease in Exome Data. Circulation Genomic and Precision Medicine, 2018, 11, e001978.	1.6	65
1176	A Distinct Phenotype of Eyes Shut Homolog (EYS)-Retinitis Pigmentosa Is Associated With Variants Near the C-Terminus. American Journal of Ophthalmology, 2018, 190, 99-112.	1.7	23
1177	Genome-wide analyses using UK Biobank data provide insights into the genetic architecture of osteoarthritis. Nature Genetics, 2018, 50, 549-558.	9.4	223
1178	Germline Lysine-Specific Demethylase 1 (LSD1/KDM1A) Mutations Confer Susceptibility to Multiple Myeloma. Cancer Research, 2018, 78, 2747-2759.	0.4	56
1179	Rare nonsynonymous variants in SORT1 are associated with increased risk for frontotemporal dementia. Neurobiology of Aging, 2018, 66, 181.e3-181.e10.	1.5	19
1180	De novo mutations in regulatory elements in neurodevelopmental disorders. Nature, 2018, 555, 611-616.	13.7	232
1181	MSeqDR mvTool: A mitochondrial DNA Web and API resource for comprehensive variant annotation, universal nomenclature collation, and reference genome conversion. Human Mutation, 2018, 39, 806-810.	1.1	32
1182	Identification of a rare BMP pathway mutation in a non-syndromic human brain arteriovenous malformation via exome sequencing. Human Genome Variation, 2018, 5, 18001.	0.4	20
1183	A point mutation in the pre-ZRS disrupts sonic hedgehog expression in the limb bud and results in triphalangeal thumbâ€“polysyndactyly syndrome. Genetics in Medicine, 2018, 20, 1405-1413.	1.1	21

#	Article	IF	Citations
1184	Biomedical informatics and machine learning for clinical genomics. Human Molecular Genetics, 2018, 27, R29-R34.	1.4	28
1185	Advanced Whole-Genome Sequencing and Analysis of Fetal Genomes from Amniotic Fluid. Clinical Chemistry, 2018, 64, 715-725.	1.5	10
1186	Prediction and interpretation of deleterious coding variants in terms of protein structural stability. Scientific Reports, 2018, 8, 4480.	1.6	64
1187	Defective kinesin binding of TUBB2A causes progressive spastic ataxia syndrome resembling sacsinopathy. Human Molecular Genetics, 2018, 27, 1892-1904.	1.4	29
1188	Mutations in the fourth Î²-propeller domain of LRP4 are associated with isolated syndactyly with fusion of the third and fourth fingers. Human Mutation, 2018, 39, 811-815.	1.1	17
1189	Report of three cases with hereditary spastic paraplegia and investigation of the mutations. Meta Gene, 2018, 16, 105-107.	0.3	1
1190	De novo and inherited mutations in the X-linked gene CLCN4 are associated with syndromic intellectual disability and behavior and seizure disorders in males and females. Molecular Psychiatry, 2018, 23, 222-230.	4.1	45
1191	The Discovery of Mutated Driver Pathways in Cancer: Models and Algorithms. IEEE/ACM Transactions on Computational Biology and Bioinformatics, 2018, 15, 988-998.	1.9	53
1192	CASP9 germline mutation in a family with multiple brain tumors. Brain Pathology, 2018, 28, 94-102.	2.1	11
1193	Deleterious proteinâ€altering mutations in the <scp>SCN10A</scp> voltageâ€gated sodium channel gene are associated with prolonged <scp>QT</scp>. Clinical Genetics, 2018, 93, 741-751.	1.0	9
1194	Identification of putative second genetic hits in schizophrenia carriers of high-risk copy number variants and resequencing in additional samples. European Archives of Psychiatry and Clinical Neuroscience, 2018, 268, 585-592.	1.8	6
1195	The Arc Gene Confers Genetic Susceptibility to Alzheimerâ€™s Disease in Han Chinese. Molecular Neurobiology, 2018, 55, 1217-1226.	1.9	30
1196	Functional variants in the sucraseâ€“isomaltase gene associate with increased risk of irritable bowel syndrome. Gut, 2018, 67, 263-270.	6.1	120
1197	Beneficial effect of ustekinumab in familial pityriasis rubra pilaris with a new missense mutation in CARD14. British Journal of Dermatology, 2018, 178, 969-972.	1.4	38
1198	A Clinical and Molecular Genetic Study of 50 Families with Autosomal Recessive Parkinsonism Revealed Known and Novel Gene Mutations. Molecular Neurobiology, 2018, 55, 3477-3489.	1.9	67
1199	The genetics of platelet count and volume in humans. Platelets, 2018, 29, 125-130.	1.1	44
1200	Resequencing Epithelial Sodium Channel Genes Identifies Rare Variants Associated With Blood Pressure Salt-Sensitivity: The GenSalt Study. American Journal of Hypertension, 2018, 31, 205-211.	1.0	25
1201	Chromatin accessibility prediction via a hybrid deep convolutional neural network. Bioinformatics, 2018, 34, 732-738.	1.8	65

#	Article	IF	Citations
1202	HEDD: Human Enhancer Disease Database. Nucleic Acids Research, 2018, 46, D113-D120.	6.5	47
1203	A novel mutation of the EYA4 gene associated with post-lingual hearing loss in a proband is co-segregating with a novel PAX3 mutation in two congenitally deaf family members. International Journal of Pediatric Otorhinolaryngology, 2018, 104, 88-93.	0.4	12
1204	Targeted massively parallel sequencing characterises the mutation spectrum of PALB2 in breast and ovarian cancer cases from Poland and Ukraine. Familial Cancer, 2018, 17, 345-349.	0.9	7
1205	FATHMM-XF: accurate prediction of pathogenic point mutations via extended features. Bioinformatics, 2018, 34, 511-513.	1.8	296
1206	Genomic analysis of the origins and evolution of multicentric diffuse lower-grade gliomas. Neuro-Oncology, 2018, 20, 632-641.	0.6	33
1207	A de novo mutation in RPL10 causes a rare X-linked ribosomopathy characterized by syndromic intellectual disability and epilepsy: A new case and review of the literature. European Journal of Medical Genetics, 2018, 61, 89-93.	0.7	22
1208	Expanding the genetic architecture and phenotypic spectrum in the skeletal ciliopathies. Human Mutation, 2018, 39, 152-166.	1.1	92
1209	Human gene essentiality. Nature Reviews Genetics, 2018, 19, 51-62.	7.7	213
1210	Genetic predisposition in children with cancer â€“ affected families' acceptance of Trio-WES. European Journal of Pediatrics, 2018, 177, 53-60.	1.3	30
1211	Insights From Molecular Characterization of Adult Patients of Families With Multigenerational Diabetes. Diabetes, 2018, 67, 137-145.	0.3	23
1212	Exome sequencing reveals NAA15 and PUF60 as candidate genes associated with intellectual disability. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 10-20.	1.1	32
1213	Biallelic mutations in <scp>DYNC2LI1</scp> are a rare cause of Ellisâ€van Creveld syndrome. Clinical Genetics, 2018, 93, 632-639.	1.0	23
1214	Two Novel Pathogenic MID1 Variants and Genotype-Phenotype Correlation Reanalysis in X-Linked Opitz G/BBB Syndrome. Molecular Syndromology, 2018, 9, 45-51.	0.3	3
1215	Interpreting the clinical significance of combined variants in multiple recessive disease genes: systematic investigation of Joubert syndrome yields little support for oligogenicity. Genetics in Medicine, 2018, 20, 223-233.	1.1	22
1216	Combined immunodeficiency and atopy caused by a dominant negative mutation in caspase activation and recruitment domain family member 11 (CARD11). Journal of Allergy and Clinical Immunology, 2018, 141, 1818-1830.e2.	1.5	93
1217	Neonatal epileptic encephalopathy caused by de novo GNAO1 mutation misdiagnosed as atypical Rett syndrome: Cautions in interpretation of genomic test results. Seminars in Pediatric Neurology, 2018, 26, 28-32.	1.0	10
1218	Clinical and genetic analysis of a rare syndrome associated with neoteny. Genetics in Medicine, 2018, 20, 495-502.	1.1	2
1219	Targeted exome sequencing in anti-factor H antibody negative HUS reveals multiple variations. Clinical and Experimental Nephrology, 2018, 22, 653-660.	0.7	21

#	Article	IF	Citations
1220	Malignant Hyperthermia in the Post-Genomics Era. Anesthesiology, 2018, 128, 168-180.	1.3	120
1221	Novel spondyloepimetaphyseal dysplasia due to UFSP2 gene mutation. Clinical Genetics, 2018, 93, 671-674.	1.0	26
1222	The role of cadherin genes in five major psychiatric disorders: A literature update. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 168-180.	1.1	45
1223	NEK1 genetic variability in a Belgian cohort of ALS and ALS-FTD patients. Neurobiology of Aging, 2018, 61, 255.e1-255.e7.	1.5	32
1224	Investigating CCNF mutations in a Taiwanese cohort with amyotrophic lateral sclerosis. Neurobiology of Aging, 2018, 62, 243.e1-243.e6.	1.5	13
1225	OPTN p.Met468Arg and ATXN2 intermediate length polyQ extension in families with C9orf72 mediated amyotrophic lateral sclerosis and frontotemporal dementia. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 75-85.	1.1	12
1226	Variants in chondroitin sulfate metabolism genes in thrombotic storm. Thrombosis Research, 2018, 161, 43-51.	0.8	5
1227	Impact of Receiving Secondary Results from Genomic Research: A 12â€Month Longitudinal Study. Journal of Genetic Counseling, 2018, 27, 709-722.	0.9	26
1228	Novel biallelic TRNT1 mutations resulting in sideroblastic anemia, combined B and T cell defects, hypogammaglobulinemia, recurrent infections, hypertrophic cardiomyopathy and developmental delay. Clinical Immunology, 2018, 188, 20-22.	1.4	24
1229	Evaluation of exome filtering techniques for the analysis of clinically relevant genes. Human Mutation, 2018, 39, 197-201.	1.1	13
1230	A novel PRRT2 pathogenic variant in a family with paroxysmal kinesigenic dyskinesia and benign familial infantile seizures. Journal of Physical Education and Sports Management, 2018, 4, a002287.	0.5	9
1231	Deleterious variants in <scp>DCHS</scp>1 are prevalent in sporadic cases of mitral valve prolapse. Molecular Genetics & Genomic Medicine, 2018, 6, 114-120.	0.6	9
1232	Redefining the phenotypic spectrum of de novo heterozygous CDK13 variants: Three patients without cardiac defects. European Journal of Medical Genetics, 2018, 61, 243-247.	0.7	14
1233	Congenital embryonal rhabdomyosarcoma caused by heterozygous concomitant PTCH1 and PTCH2 germline mutations. European Journal of Human Genetics, 2018, 26, 137-142.	1.4	17
1234	Evaluation of reported pathogenic variants and their frequencies in a Japanese population based on a whole-genome reference panel of 2049 individuals. Journal of Human Genetics, 2018, 63, 213-230.	1.1	35
1235	Properties of human genes guided by their enrichment in rare and common variants. Human Mutation, 2018, 39, 365-370.	1.1	13
1236	Clinical and genetic characterization of AP4B1â€associated SPG47. American Journal of Medical Genetics, Part A, 2018, 176, 311-318.	0.7	47
1237	Relaxed Selection During a Recent Human Expansion. Genetics, 2018, 208, 763-777.	1.2	49

#	Article	IF	Citations
1238	A craniosynostosis massively parallel sequencing panel study in 309 Australian and New Zealand patients: findings and recommendations. Genetics in Medicine, 2018, 20, 1061-1068.	1.1	37
1239	ERCC4 variants identified in a cohort of patients with segmental progeroid syndromes. Human Mutation, 2018, 39, 255-265.	1.1	23
1240	Understanding Neurodevelopmental Disorders: The Promise of Regulatory Variation in the 3â€²UTRome. Biological Psychiatry, 2018, 83, 548-557.	0.7	48
1241	Tarsalâ€carpal coalition syndrome: Report of a novel missense mutation in NOG gene and phenotypic delineation. American Journal of Medical Genetics, Part A, 2018, 176, 219-224.	0.7	12
1242	Exome Sequencing Identifies LOXL2 Mutation as a Cause of Familial Intracranial Aneurysm. World Neurosurgery, 2018, 109, e812-e818.	0.7	22
1243	EYSmutation update: In silico assessment of 271 reported and 26 novel variants in patients with retinitis pigmentosa. Human Mutation, 2018, 39, 177-186.	1.1	23
1244	Molecular and functional characterization of familial chylomicronemia syndrome. Atherosclerosis, 2018, 269, 272-278.	0.4	16
1245	Identification of genes carrying rare variants of moderate to large effect in schizophrenia: A replication study. Schizophrenia Research, 2018, 197, 577-578.	1.1	0
1246	Identification of de novo germline mutations and causal genes for sporadic diseases using trioâ€based wholeâ€exome/genome sequencing. Biological Reviews, 2018, 93, 1014-1031.	4.7	35
1247	Biallelic interferon regulatory factor 8 mutation: AÂcomplex immunodeficiency syndrome with dendritic cell deficiency, monocytopenia, and immune dysregulation. Journal of Allergy and Clinical Immunology, 2018, 141, 2234-2248.	1.5	63
1248	Biallelic mutations in mitochondrial tryptophanylâ€tRNA synthetase cause Levodopaâ€responsive infantileâ€onset Parkinsonism. Clinical Genetics, 2018, 93, 712-718.	1.0	31
1249	Dominant-negative loss of function arises from a second, more frequent variant within the SAND domain of autoimmune regulator (AIRE). Journal of Autoimmunity, 2018, 88, 114-120.	3.0	29
1250	Homozygosity for a nonsense variant in AIMP2 is associated with a progressive neurodevelopmental disorder with microcephaly, seizures, and spastic quadriparesis. Journal of Human Genetics, 2018, 63, 19-25.	1.1	26
1251	Genetic Costs of Domestication and Improvement. Journal of Heredity, 2018, 109, 103-116.	1.0	149
1252	The genetic basis for most patients with pustular skin disease remains elusive. British Journal of Dermatology, 2018, 178, 740-748.	1.4	82
1253	Hereditary angioedema with a mutation in the plasminogen gene. Allergy: European Journal of Allergy and Clinical Immunology, 2018, 73, 442-450.	2.7	225
1254	SMART Cancer Navigator: A Framework for Implementing ASCO Workshop Recommendations to Enable Precision Cancer Medicine. JCO Precision Oncology, 2018, 2018, 1-14.	1.5	19
1255	A Subset of Mesotheliomas With Improved Survival Occurring in Carriers of BAP1 and Other Germline Mutations. Journal of Clinical Oncology, 2018, 36, 3485-3494.	0.8	104

#	Article	IF	Citations
1256	Artificial Intelligence Approach for Variant Reporting. JCO Clinical Cancer Informatics, 2018, 2, 1-13.	1.0	13
1257	TP53 Germline Variations Influence the Predisposition and Prognosis of B-Cell Acute Lymphoblastic Leukemia in Children. Journal of Clinical Oncology, 2018, 36, 591-599.	0.8	121
1258	Missense variants in <scp>TMEM</scp>67 in a patient with Joubert syndrome. Clinical Case Reports (discontinued), 2018, 6, 2189-2192.	0.2	3
1259	iMEGES: integrated mental-disorder GEnome score by deep neural network for prioritizing the susceptibility genes for mental disorders in personal genomes. BMC Bioinformatics, 2018, 19, 501.	1.2	10
1260	Identifying SYNE1 Ataxia With Novel Mutations in a Chinese Population. Frontiers in Neurology, 2018, 9, 1111.	1.1	14
1261	Ensembl variation resources. Database: the Journal of Biological Databases and Curation, 2018, 2018, .	1.4	377
1262	Representativeness of variation benchmark datasets. BMC Bioinformatics, 2018, 19, 461.	1.2	18
1263	GRIPT: a novel case-control analysis method for Mendelian disease gene discovery. Genome Biology, 2018, 19, 203.	3.8	3
1264	Rare genetic variation implicated in non-Hispanic white families with Alzheimer disease. Neurology: Genetics, 2018, 4, e286.	0.9	27
1265	Targeted Gene Sequencing in Children with Crohnâ€™s Disease and Their Parents: Implications for Missing Heritability. G3: Genes, Genomes, Genetics, 2018, 8, 2881-2888.	0.8	1
1266	Towards a Central Role of ISL1 in the Bladder Exstrophyâ€“Epispadias Complex (BEEC): Computational Characterization of Genetic Variants and Structural Modelling. Genes, 2018, 9, 609.	1.0	6
1267	Combinatorial Approach for Complex Disorder Prediction: Case Study of Neurodevelopmental Disorders. Genetics, 2018, 210, 1483-1495.	1.2	1
1268	De novo variants in congenital diaphragmatic hernia identify MYRF as a new syndrome and reveal genetic overlaps with other developmental disorders. PLoS Genetics, 2018, 14, e1007822.	1.5	79
1269	Phosphorylation of Parkin at serine 65 is essential for its activationin vivo. Open Biology, 2018, 8, 180108.	1.5	81
1270	Imputation-Aware Tag SNP Selection To Improve Power for Large-Scale, Multi-ethnic Association Studies. G3: Genes, Genomes, Genetics, 2018, 8, 3255-3267.	0.8	36
1271	Assessment of coding region variants in Kuwaiti population: implications for medical genetics and population genomics. Scientific Reports, 2018, 8, 16583.	1.6	26
1272	Cohort-wide deep whole genome sequencing and the allelic architecture of complex traits. Nature Communications, 2018, 9, 4674.	5.8	33
1273	Mutations in MAST1 Cause Mega-Corpus-Callosum Syndrome with Cerebellar Hypoplasia and Cortical Malformations. Neuron, 2018, 100, 1354-1368.e5.	3.8	35

#	Article	IF	Citations
1274	Functional characterization of TBR1 variants in neurodevelopmental disorder. Scientific Reports, 2018, 8, 14279.	1.6	26
1275	Identification of novel LEPR mutations in Pakistani families with morbid childhood obesity. BMC Medical Genetics, 2018, 19, 199.	2.1	8
1276	Cell-Free DNA From Metastatic Pancreatic Neuroendocrine Tumor Patients Contains Tumor-Specific Mutations and Copy Number Variations. Frontiers in Oncology, 2018, 8, 467.	1.3	25
1277	Ablepharon and craniosynostosis in a patient with a localized TWIST1 basic domain substitution. American Journal of Medical Genetics, Part A, 2018, 176, 2777-2780.	0.7	7
1278	SeqVItA: Sequence Variant Identification and Annotation Platform for Next Generation Sequencing Data. Frontiers in Genetics, 2018, 9, 537.	1.1	4
1279	A de novo pathogenic CSNK1E mutation identified by exome sequencing in family trios with epileptic encephalopathy. Human Mutation, 2019, 40, 281-287.	1.1	7
1280	Pathogenic Variants in Fucokinase Cause a Congenital Disorder of Glycosylation. American Journal of Human Genetics, 2018, 103, 1030-1037.	2.6	18
1281	Spastic paraplegia due to SPAST mutations is modified by the underlying mutation and sex. Brain, 2018, 141, 3331-3342.	3.7	72
1282	<scp>TUBB</scp> 1 mutations cause thyroid dysgenesis associated with abnormal platelet physiology. EMBO Molecular Medicine, 2018, 10, .	3.3	47
1283	Non-Coding Variants in BRCA1 and BRCA2 Genes: Potential Impact on Breast and Ovarian Cancer Predisposition. Cancers, 2018, 10, 453.	1.7	14
1284	Mutational and phenotypic spectra of KCNE1 deficiency in Jervell and Langeâ€Nielsen Syndrome and Romanoâ€Ward Syndrome. Human Mutation, 2019, 40, 162-176.	1.1	44
1285	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	3.5	86
1286	A Young Male with Parafibromin-Deficient Parathyroid Carcinoma Due to a Rare Germline HRPT2/CDC73 Mutation. Endocrine Pathology, 2018, 29, 374-379.	5.2	6
1287	Functional characterization of TRPM4 variants identified in sudden unexpected natural death. Forensic Science International, 2018, 293, 37-46.	1.3	11
1288	Neomorphic PDGFRA extracellular domain driver mutations are resistant to PDGFRA targeted therapies. Nature Communications, 2018, 9, 4583.	5.8	44
1289	Delineation of Novel Compound Heterozygous Variants in LTBP2 Associated with Juvenile Open Angle Glaucoma. Genes, 2018, 9, 527.	1.0	17
1290	A novel PNPLA6 compound heterozygous mutation identified in a Chinese patient with Boucherâ€'NeuhÃ¯Â¿Â½user syndrome. Molecular Medicine Reports, 2018, 18, 261-267.	1.1	14
1291	MDSGene: Closing Data Gaps in Genotype-Phenotype Correlations of Monogenic Parkinsonâ€™s Disease. Journal of Parkinson's Disease, 2018, 8, S25-S30.	1.5	33

#	Article	IF	Citations
1292	Genomic Studies in a Large Cohort of Hearing Impaired Italian Patients Revealed Several New Alleles, a Rare Case of Uniparental Disomy (UPD) and the Importance to Search for Copy Number Variations. Frontiers in Genetics, 2018, 9, 681.	1.1	25
1293	Calculating the statistical significance of rare variants causal for Mendelian and complex disorders. BMC Medical Genomics, 2018, 11, 53.	0.7	13
1294	Whole-genome sequencing in a family with twin boys with autism and intellectual disability suggests multimodal polygenic risk. Journal of Physical Education and Sports Management, 2018, 4, a003285.	0.5	11
1295	Human IFN-Î³ immunity to mycobacteria is governed by both IL-12 and IL-23. Science Immunology, 2018, 3, .	5.6	152
1296	The sequencing and interpretation of the genome obtained from a Serbian individual. PLoS ONE, 2018, 13, e0208901.	1.1	3
1297	Need for high-resolution Genetic Analysis in iPSC: Results and Lessons from the ForIPS Consortium. Scientific Reports, 2018, 8, 17201.	1.6	70
1298	Large-scale genome-wide meta-analysis of polycystic ovary syndrome suggests shared genetic architecture for different diagnosis criteria. PLoS Genetics, 2018, 14, e1007813.	1.5	341
1299	Tuberculosis and impaired IL-23â€“dependent IFN-Î³ immunity in humans homozygous for a common TYK2 missense variant. Science Immunology, 2018, 3, .	5.6	148
1300	Diagnosing rare diseases after the exome. Journal of Physical Education and Sports Management, 2018, 4, a003392.	0.5	48
1301	Personal Genome Project UK (PGP-UK): a research and citizen science hybrid project in support of personalized medicine. BMC Medical Genomics, 2018, 11, 108.	0.7	34
1302	Identification of Common and Rare Genetic Variation Associated With Plasma Protein Levels Using Whole-Exome Sequencing and Mass Spectrometry. Circulation Genomic and Precision Medicine, 2018, 11, e002170.	1.6	26
1303	Aberrant Inclusion of a Poison Exon Causes Dravet Syndrome and Related SCN1A-Associated Genetic Epilepsies. American Journal of Human Genetics, 2018, 103, 1022-1029.	2.6	76
1304	Neutropenia in the age of genetic testing: Advances and challenges. American Journal of Hematology, 2019, 94, 384-393.	2.0	18
1305	OligoPVP: Phenotype-driven analysis of individual genomic information to prioritize oligogenic disease variants. Scientific Reports, 2018, 8, 14681.	1.6	8
1306	The Genetic Landscape of Diamond-Blackfan Anemia. American Journal of Human Genetics, 2018, 103, 930-947.	2.6	184
1307	Challenging popular tools for the annotation of genetic variations with a real case, pathogenic mutations of lysosomal alpha-galactosidase. BMC Bioinformatics, 2018, 19, 433.	1.2	8
1308	De novo mutations in the GTP/GDP-binding region of RALA, a RAS-like small GTPase, cause intellectual disability and developmental delay. PLoS Genetics, 2018, 14, e1007671.	1.5	16
1309	The E3ÂUbiquitin Ligases TRIM17 and TRIM41 Modulate Î±-Synuclein Expression by Regulating ZSCAN21. Cell Reports, 2018, 25, 2484-2496.e9.	2.9	34

#	Article	IF	Citations
1310	Combined CNV, haplotyping and whole exome sequencing enable identification of two distinct novel EYS mutations causing RP in a single inbred tribe. American Journal of Medical Genetics, Part A, 2018, 176, 2695-2703.	0.7	3
1311	A semi-supervised approach for predicting cell-type specific functional consequences of non-coding variation using MPRAs. Nature Communications, 2018, 9, 5199.	5.8	34
1312	Searching for Essential Genes in the Cancer Genome. Biochemistry (Moscow) Supplement Series B: Biomedical Chemistry, 2018, 12, 283-296.	0.2	0
1313	Variable Clinical Characteristics and Molecular Spectrum of Patients with Syndromes of Reduced Sensitivity to Thyroid Hormone: Genetic Defects in the <i>THRB</i> and <i>SLC16A2</i> Genes. Hormone Research in Paediatrics, 2018, 90, 283-290.	0.8	10
1314	Computational Methods for the Pharmacogenetic Interpretation of Next Generation Sequencing Data. Frontiers in Pharmacology, 2018, 9, 1437.	1.6	62
1315	A biallelic truncating AEBP1 variant causes connective tissue disorder in two siblings. American Journal of Medical Genetics, Part A, 2019, 179, 50-56.	0.7	11
1316	Haploinsufficiency of NCOR1 associated with autism spectrum disorder, scoliosis, and abnormal palatogenesis. American Journal of Medical Genetics, Part A, 2018, 176, 2466-2469.	0.7	5
1317	Human genetic variation in GLS2 is associated with development of complicated Staphylococcus aureus bacteremia. PLoS Genetics, 2018, 14, e1007667.	1.5	16
1318	Functional architecture of low-frequency variants highlights strength of negative selection across coding and non-coding annotations. Nature Genetics, 2018, 50, 1600-1607.	9.4	132
1319	Burden Testing of Rare Variants Identified through Exome Sequencing via Publicly Available Control Data. American Journal of Human Genetics, 2018, 103, 522-534.	2.6	132
1320	T-cell defects in patients with ARPC1B germline mutations account for combined immunodeficiency. Blood, 2018, 132, 2362-2374.	0.6	99
1321	Genome-wide association study of 23,500 individuals identifies 7 loci associated with brain ventricular volume. Nature Communications, 2018, 9, 3945.	5.8	31
1322	Genome-wide meta-analysis of 158,000 individuals of European ancestry identifies three loci associated with chronic back pain. PLoS Genetics, 2018, 14, e1007601.	1.5	112
1323	Exome scale map of genetic alterations promoting metastasis in colorectal cancer. BMC Genetics, 2018, 19, 85.	2.7	22
1324	The Clinical Cases of Geleophysic Dysplasia: One Gene, Different Phenotypes. Case Reports in Endocrinology, 2018, 2018, 1-7.	0.2	9
1325	Bi-allelic Mutations in NDUFA6 Establish Its Role in Early-Onset Isolated Mitochondrial Complex I Deficiency. American Journal of Human Genetics, 2018, 103, 592-601.	2.6	41
1326	Penetrance and Expressivity in Inherited Cancer Predisposing Syndromes. Trends in Cancer, 2018, 4, 718-728.	3.8	36
1327	A novel nonsense autosomal dominant mutation in the GLRA1 gene causing hyperekplexia. Journal of Neural Transmission, 2018, 125, 1877-1883.	1.4	4

#	Article	IF	Citations
1328	NBEA: Developmental disease gene with early generalized epilepsy phenotypes. Annals of Neurology, 2018, 84, 788-795.	2.8	44
1329	Early-Onset Invasive Infection Due to Corynespora cassiicola Associated with Compound Heterozygous CARD9 Mutations in a Colombian Patient. Journal of Clinical Immunology, 2018, 38, 794-803.	2.0	40
1330	Genetic Variants in SGLT1, Glucose Tolerance, and Cardiometabolic Risk. Journal of the American College of Cardiology, 2018, 72, 1763-1773.	1.2	61
1331	Metaâ€Analysis of Genetic Influences on Initial Alcohol Sensitivity. Alcoholism: Clinical and Experimental Research, 2018, 42, 2349-2359.	1.4	21
1332	Replication of a rare risk haplotype on 1p36.33 for autism spectrum disorder. Human Genetics, 2018, 137, 807-815.	1.8	4
1333	Exhaustive non-synonymous variants functionality prediction enables high resolution characterization of the neurofibromin architecture. EBioMedicine, 2018, 36, 508-516.	2.7	1
1334	9p24 triplication in syndromic hydrocephalus with diffuse villous hyperplasia of the choroid plexus. Journal of Physical Education and Sports Management, 2018, 4, a003145.	0.5	8
1335	Whole-Genome Linkage Scan Combined With Exome Sequencing Identifies Novel Candidate Genes for Carotid Intima-Media Thickness. Frontiers in Genetics, 2018, 9, 420.	1.1	3
1336	Mutations in KCNK4 that Affect Gating Cause a Recognizable Neurodevelopmental Syndrome. American Journal of Human Genetics, 2018, 103, 621-630.	2.6	73
1337	GRIN2D variants in three cases of developmental and epileptic encephalopathy. Clinical Genetics, 2018, 94, 538-547.	1.0	17
1338	Deep generative models of genetic variation capture the effects of mutations. Nature Methods, 2018, 15, 816-822.	9.0	395
1339	Genome-wide association studies and CRISPR/Cas9-mediated gene editing identify regulatory variants influencing eyebrow thickness in humans. PLoS Genetics, 2018, 14, e1007640.	1.5	20
1340	ENTPRISE-X: Predicting disease-associated frameshift and nonsense mutations. PLoS ONE, 2018, 13, e0196849.	1.1	20
1341	Exome-Wide Rare Variant Analyses in Sudden Infant Death Syndrome. Journal of Pediatrics, 2018, 203, 423-428.e11.	0.9	17
1342	Machine Learning Classification and Structureâ€“Functional Analysis of Cancer Mutations Reveal Unique Dynamic and Network Signatures of Driver Sites in Oncogenes and Tumor Suppressor Genes. Journal of Chemical Information and Modeling, 2018, 58, 2131-2150.	2.5	20
1343	Mutated Thyroid Hormone Transporter OATP1C1 Associates with Severe Brain Hypometabolism and Juvenile Neurodegeneration. Thyroid, 2018, 28, 1406-1415.	2.4	57
1344	De novo Mutations (DNMs) in Autism Spectrum Disorder (ASD): Pathway and Network Analysis. Frontiers in Genetics, 2018, 9, 406.	1.1	40
1345	TIVAN: tissue-specific cis-eQTL single nucleotide variant annotation and prediction. Bioinformatics, 2019, 35, 1573-1575.	1.8	14

#	Article	IF	Citations
1347	Prioritization of Variants Detected by Next Generation Sequencing According to the Mutation Tolerance and Mutational Architecture of the Corresponding Genes. International Journal of Molecular Sciences, 2018, 19, 1584.	1.8	16
1348	Common Variable Immunodeficiency with Genetic Defects Identified by Whole Exome Sequencing. BioMed Research International, 2018, 2018, 1-7.	0.9	18
1349	Defining the molecular signatures of Achilles tendinopathy and anterior cruciate ligament ruptures: A whole-exome sequencing approach. PLoS ONE, 2018, 13, e0205860.	1.1	16
1351	CRISPR-Cas9 human gene replacement and phenomic characterization in Caenorhabditis elegans to understand the functional conservation of human genes and decipher variants of uncertain significance. DMM Disease Models and Mechanisms, 2018, 11, .	1.2	38
1352	Multiplexed Targeted Resequencing Identifies Coding and Regulatory Variation Underlying Phenotypic Extremes of High-Density Lipoprotein Cholesterol in Humans. Circulation Genomic and Precision Medicine, 2018, 11, e002070.	1.6	5
1353	PINES: phenotype-informed tissue weighting improves prediction of pathogenic noncoding variants. Genome Biology, 2018, 19, 173.	3.8	28
1354	Interpretation of the noncoding genome in medicine. Personalized Medicine, 2018, 15, 453-455.	0.8	2
1355	Impact of structural prior knowledge in SNV prediction: Towards causal variant finding in rare disease. PLoS ONE, 2018, 13, e0204101.	1.1	3
1356	Genomic analysis of DNA repair genes and androgen signaling in prostate cancer. BMC Cancer, 2018, 18, 960.	1.1	55
1357	Bioinformatics: Sequences, Structures, Phylogeny. , 2018, , .		0
1358	Large-scale whole-exome sequencing association studies identify rare functional variants influencing serum urate levels. Nature Communications, 2018, 9, 4228.	5.8	43
1359	Predicting variant deleteriousness in non-human species: applying the CADD approach in mouse. BMC Bioinformatics, 2018, 19, 373.	1.2	10
1360	A novel PAX3 mutation in a Korean patient with Waardenburg syndrome type 1 and unilateral branch retinal vein and artery occlusion: a case report. BMC Ophthalmology, 2018, 18, 266.	0.6	9
1361	Discovery of potential causative mutations in human coding and noncoding genome with the interactive software BasePlayer. Nature Protocols, 2018, 13, 2580-2600.	5.5	27
1362	Mis-splicing of the GALNS gene resulting from deep intronic mutations as a cause of Morquio a disease. BMC Medical Genetics, 2018, 19, 183.	2.1	14
1363	Syndromic hearing loss molecular diagnosis: Application of massive parallel sequencing. Hearing Research, 2018, 370, 181-188.	0.9	6
1364	Bioinformatics in Clinical Genomic Sequencing. Advances in Molecular Pathology, 2018, 1, 9-26.	0.2	1
1365	Discovering Genetic Factors for psoriasis through exhaustively searching for significant second order SNP-SNP interactions. Scientific Reports, 2018, 8, 15186.	1.6	16

#	Article	IF	Citations
1366	Computational Epigenomics and Its Application in Regulatory Genomics. , 2018, , 115-139.		0
1367	Characterization of Nigerian breast cancer reveals prevalent homologous recombination deficiency and aggressive molecular features. Nature Communications, 2018, 9, 4181.	5.8	77
1368	Whole Exome Sequencing of HIV-1 long-term non-progressors identifies rare variants in genes encoding innate immune sensors and signaling molecules. Scientific Reports, 2018, 8, 15253.	1.6	12
1369	A unique de novo gain-of-function variant inCAMK4associated with intellectual disability and hyperkinetic movement disorder. Journal of Physical Education and Sports Management, 2018, 4, a003293.	0.5	16
1370	Rho Guanine Nucleotide Exchange Factor ARHGEF17 Is a Risk Gene for Intracranial Aneurysms. Circulation Genomic and Precision Medicine, 2018, 11, e002099.	1.6	18
1371	Sudden unexpected death in the young â€" Value of massive parallel sequencing in postmortem genetic analyses. Forensic Science International, 2018, 293, 70-76.	1.3	17
1372	Deciphering the Emerging Complexities of Molecular Mechanisms at GWAS Loci. American Journal of Human Genetics, 2018, 103, 637-653.	2.6	93
1373	Biallelic Mutations in LRRC56, Encoding a Protein Associated with Intraflagellar Transport, Cause Mucociliary Clearance and Laterality Defects. American Journal of Human Genetics, 2018, 103, 727-739.	2.6	49
1374	Plasticity-related gene 3 (LPPR1) and age at diagnosis of Parkinson disease. Neurology: Genetics, 2018, 4, e271.	0.9	12
1375	N-Acetylglutamate Synthase Deficiency Due to a Recurrent Sequence Variant in the N-acetylglutamate Synthase Enhancer Region. Scientific Reports, 2018, 8, 15436.	1.6	7
1376	Guidelineâ€based and bioinformatic reassessment of lesionâ€associated gene and variant pathogenicity in focal human epilepsies. Epilepsia, 2018, 59, 2145-2152.	2.6	8
1377	A Bayesian framework for efficient and accurate variant prediction. PLoS ONE, 2018, 13, e0203553.	1.1	12
1378	Extremely rare variants reveal patterns of germline mutation rate heterogeneity in humans. Nature Communications, 2018, 9, 3753.	5.8	121
1379	Genetic screening for macular dystrophies in patients clinically diagnosed with dry ageâ€related macular degeneration. Clinical Genetics, 2018, 94, 569-574.	1.0	18
1380	Exome-wide analysis of bi-allelic alterations identifies a Lynch phenotype in The Cancer Genome Atlas. Genome Medicine, 2018, 10, 69.	3.6	10
1381	Extracting complementary insights from molecular phenotypes for prioritization of disease-associated mutations. Current Opinion in Systems Biology, 2018, 11, 107-116.	1.3	4
1382	Exome sequencing-based identification of novel type 2 diabetes risk allele loci in the Qatari population. PLoS ONE, 2018, 13, e0199837.	1.1	7
1383	A novel panel of short mononucleotide repeats linked to informative polymorphisms enabling effective high volume low cost discrimination between mismatch repair deficient and proficient tumours. PLoS ONE, 2018, 13, e0203052.	1.1	14

#	Article	IF	Citations
1384	Synergistic Interaction of HOXB13 and CIP2A Predisposes to Aggressive Prostate Cancer. Clinical Cancer Research, 2018, 24, 6265-6276.	3.2	17
1385	Complex Haplotypes of GSTM1 Gene Deletions Harbor Signatures of a Selective Sweep in East Asian Populations. G3: Genes, Genomes, Genetics, 2018, 8, 2953-2966.	0.8	8
1386	Assessing Human Genetic Variations in Glucose Transporter SLC2A10 and Their Role in Altering Structural and Functional Properties. Frontiers in Genetics, 2018, 9, 276.	1.1	12
1387	Genomic screening in rare disorders: New mutations and phenotypes, highlighting ALG14 as a novel cause of severe intellectual disability. Clinical Genetics, 2018, 94, 528-537.	1.0	29
1388	Variants in cardiac <scp>GATA</scp> genes associated with bicuspid aortic valve. European Journal of Clinical Investigation, 2018, 48, e13027.	1.7	13
1389	Large-scale meta-analysis highlights the hypothalamicâ€“pituitaryâ€“gonadal axis in the genetic regulation of menstrual cycle length. Human Molecular Genetics, 2018, 27, 4323-4332.	1.4	20
1390	<scp>OXA</scp> 1L mutations cause mitochondrial encephalopathy and a combined oxidative phosphorylation defect. EMBO Molecular Medicine, 2018, 10, .	3.3	54
1391	Accurate classification of BRCA1 variants with saturation genome editing. Nature, 2018, 562, 217-222.	13.7	570
1392	Comparative Genomics Approaches Accurately Predict Deleterious Variants in Plants. G3: Genes, Genomes, Genetics, 2018, 8, 3321-3329.	0.8	36
1393	ClinPred: Prediction Tool to Identify Disease-Relevant Nonsynonymous Single-Nucleotide Variants. American Journal of Human Genetics, 2018, 103, 474-483.	2.6	149
1394	Genome-wide association meta-analysis of coronary artery disease and periodontitis reveals a novel shared risk locus. Scientific Reports, 2018, 8, 13678.	1.6	35
1395	Confirmation of the Role ofDHX38in the Etiology of Early-Onset Retinitis Pigmentosa. , 2018, 59, 4552.		16
1396	Varicella-zoster virus CNS vasculitis and RNA polymerase III gene mutation in identical twins. Neurology: Neuroimmunology and NeuroInflammation, 2018, 5, e500.	3.1	49
1397	Identification of a pathogenic PMP2 variant in a multi-generational family with CMT type 1: Clinical gene panels versus genome-wide approaches to molecular diagnosis. Molecular Genetics and Metabolism, 2018, 125, 302-304.	0.5	13
1398	Case Report: Identification of an HNF1B p.Arg527Gln mutation in a Maltese patient with atypical early onset diabetes and diabetic nephropathy. BMC Endocrine Disorders, 2018, 18, 28.	0.9	11
1399	No major role for rare plectin variants in arrhythmogenic right ventricular cardiomyopathy. PLoS ONE, 2018, 13, e0203078.	1.1	2
1400	Novel missense and 3â€²-UTR splice site variants in LHFPL5 cause autosomal recessive nonsyndromic hearing impairment. Journal of Human Genetics, 2018, 63, 1099-1107.	1.1	3
1401	Whole Exome Sequencing Identifies New Host Genomic Susceptibility Factors in Empyema Caused by Streptococcus pneumoniae in Children: A Pilot Study. Genes, 2018, 9, 240.	1.0	9

#	Article	IF	Citations
1402	A novel association of campomelic dysplasia and hydrocephalus with an unbalanced chromosomal translocation upstream of SOX9. Journal of Physical Education and Sports Management, 2018, 4, a002766.	0.5	8
1403	Variable cardiovascular phenotypes associated with SMAD2 pathogenic variants. Human Mutation, 2018, 39, 1875-1884.	1.1	23
1404	Rare coding variant analysis in a large cohort of Ashkenazi Jewish families with inflammatory bowel disease. Human Genetics, 2018, 137, 723-734.	1.8	8
1405	Novel candidate genes and variants underlying autosomal recessive neurodevelopmental disorders with intellectual disability. Human Genetics, 2018, 137, 735-752.	1.8	42
1406	Multiple endocrine neoplasia type 1: clinical correlates of MEN1 gene methylation. Pathology, 2018, 50, 622-628.	0.3	7
1407	Frequent STAT3 mutations in CD8+ T cells from patients with pure red cell aplasia. Blood Advances, 2018, 2, 2704-2712.	2.5	38
1408	Identification of rare de novo epigenetic variations in congenital disorders. Nature Communications, 2018, 9, 2064.	5.8	82
1409	ActiveDriverDB: human disease mutations and genome variation in post-translational modification sites of proteins. Nucleic Acids Research, 2018, 46, D901-D910.	6.5	82
1410	A novel missense variant in the SDR domain of the WWOX gene leads to complete loss of WWOX protein with early-onset epileptic encephalopathy and severe developmental delay. Neurogenetics, 2018, 19, 151-156.	0.7	20
1411	Study of 300,486 individuals identifies 148 independent genetic loci influencing general cognitive function. Nature Communications, 2018, 9, 2098.	5.8	484
1412	From genome-wide associations to candidate causal variants by statistical fine-mapping. Nature Reviews Genetics, 2018, 19, 491-504.	7.7	611
1413	Copy number variants implicate cardiac function and development pathways in earthquake-induced stress cardiomyopathy. Scientific Reports, 2018, 8, 7548.	1.6	8
1414	Asn391Thr Mutation of Î²-Myosin Heavy Chain in a Hypertrophic Cardiomyopathy Family. International Heart Journal, 2018, 59, 596-600.	0.5	5
1415	Diagnostic Yield of Next-generation Sequencing in Very Early-onset Inflammatory Bowel Diseases: A Multicentre Study. Journal of Crohn's and Colitis, 2018, 12, 1104-1112.	0.6	68
1416	Demographic History and Genetic Adaptation in the Himalayan Region Inferred from Genome-Wide SNP Genotypes of 49 Populations. Molecular Biology and Evolution, 2018, 35, 1916-1933.	3.5	36
1417	Genetic-Driven Druggable Target Identification and Validation. Trends in Genetics, 2018, 34, 558-570.	2.9	44
1418	Naturally Occurring Missense MRGPRX2 Variants Display Loss of Function Phenotype for Mast Cell Degranulation in Response to Substance P, Hemokinin-1, Human Î²-Defensin-3, and Icatibant. Journal of Immunology, 2018, 201, 343-349.	0.4	57
1419	SimPEL: Simulationâ€based power estimation for sequencing studies of lowâ€prevalence conditions. Genetic Epidemiology, 2018, 42, 480-487.	0.6	0

#	Article	IF	Citations
1420	Functional Assays Are Essential for Interpretation of Missense Variants Associated with Variable Expressivity. American Journal of Human Genetics, 2018, 102, 1062-1077.	2.6	69
1421	Evaluating the causality of novel sequence variants in the prion protein gene by example. Neurobiology of Aging, 2018, 71, 265.e1-265.e7.	1.5	9
1422	Systematic pan-cancer analysis of somatic allele frequency. Scientific Reports, 2018, 8, 7735.	1.6	21
1423	GWAS4D: multidimensional analysis of context-specific regulatory variant for human complex diseases and traits. Nucleic Acids Research, 2018, 46, W114-W120.	6.5	69
1424	Wholeâ€exome sequencing for variant discovery in blepharospasm. Molecular Genetics & Genomic Medicine, 2018, 6, 601-626.	0.6	20
1425	De novo FBXO11 mutations are associated with intellectual disability and behavioural anomalies. Human Genetics, 2018, 137, 401-411.	1.8	29
1426	Gain-of-function KCNJ6 Mutation in a Severe Hyperkinetic Movement Disorder Phenotype. Neuroscience, 2018, 384, 152-164.	1.1	18
1427	Naturally Occurring Variants in LRP1 (Low-Density Lipoprotein Receptorâ€"Related Protein 1) Affect HDL (High-Density Lipoprotein) Metabolism Through ABCA1 (ATP-Binding Cassette A1) and SR-B1 (Scavenger) Tj ETQq1 1 0.784314 rgBT /O 1440-1453.	1.1	13
1428	Whole exome sequencing identifies PLEC, EXO5 and DNAH7 as novel susceptibility genes in testicular cancer. International Journal of Cancer, 2018, 143, 1954-1962.	2.3	19
1429	A pilot study of exome sequencing in a diverse New Zealand cohort with undiagnosed disorders and cancer. Journal of the Royal Society of New Zealand, 2018, 48, 262-279.	1.0	2
1430	SNPnexus: assessing the functional relevance of genetic variation to facilitate the promise of precision medicine. Nucleic Acids Research, 2018, 46, W109-W113.	6.5	163
1431	Exome sequencing revealed C1Q homozygous mutation in Pediatric Systemic Lupus Erythematosus. Allergologia Et Immunopathologia, 2018, 46, 594-598.	1.0	5
1432	SCN5A (Na _V 1.5) Variant Functional Perturbation and Clinical Presentation. Circulation Genomic and Precision Medicine, 2018, 11, e002095.	1.6	36
1433	Conditional mouse models support the role of SLC39A14 (ZIP14) in Hyperostosis Cranialis Interna and in bone homeostasis. PLoS Genetics, 2018, 14, e1007321.	1.5	13
1434	De Novo Mutation in Genes Regulating Neural Stem Cell Fate in Human Congenital Hydrocephalus. Neuron, 2018, 99, 302-314.e4.	3.8	112
1435	Genetic selection of athletic success in sport-hunting dogs. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E7212-E7221.	3.3	54
1436	Further evidence for the involvement of EFL1 in a Shwachmanâ€"Diamond-like syndrome and expansion of the phenotypic features. Journal of Physical Education and Sports Management, 2018, 4, a003046.	0.5	29
1437	Data Mining: Applying the AD&FTD Mutation Database to Progranulin. Methods in Molecular Biology, 2018, 1806, 81-92.	0.4	6

#	Article	IF	Citations
1438	De novo variants in GREB1L are associated with non-syndromic inner ear malformations and deafness. Human Genetics, 2018, 137, 459-470.	1.8	24
1439	PopViz: a webserver for visualizing minor allele frequencies and damage prediction scores of human genetic variations. Bioinformatics, 2018, 34, 4307-4309.	1.8	55
1440	Reply: ATAD1 encephalopathy and stiff baby syndrome: a recognizable clinical presentation. Brain, 2018, 141, e50-e50.	3.7	1
1441	GCH1 mutations in dopa-responsive dystonia and Parkinsonâ€™s disease. Journal of Neurology, 2018, 265, 1860-1870.	1.8	29
1442	Whole-Genome Sequencing in Severe Chronic Obstructive Pulmonary Disease. American Journal of Respiratory Cell and Molecular Biology, 2018, 59, 614-622.	1.4	22
1443	The genetic component of preeclampsia: A whole-exome sequencing study. PLoS ONE, 2018, 13, e0197217.	1.1	21
1444	Validation of CZECANCA (CZEch CAncer paNel for Clinical Application) for targeted NGS-based analysis of hereditary cancer syndromes. PLoS ONE, 2018, 13, e0195761.	1.1	31
1445	Meta-analysis of genome-wide association studies for neuroticism in 449,484 individuals identifies novel genetic loci and pathways. Nature Genetics, 2018, 50, 920-927.	9.4	564
1446	Genome-wide association meta-analysis in 269,867 individuals identifies new genetic and functional links to intelligence. Nature Genetics, 2018, 50, 912-919.	9.4	893
1447	A variant in LMX1A causes autosomal recessive severe-to-profound hearing impairment. Human Genetics, 2018, 137, 471-478.	1.8	18
1448	Functional characterization of biallelic RTTN variants identified in an infant with microcephaly, simplified gyral pattern, pontocerebellar hypoplasia, and seizures. Pediatric Research, 2018, 84, 435-441.	1.1	11
1449	Breast cancer in women with neurofibromatosis type 1 (NF1): a comprehensive case series with molecular insights into its aggressive phenotype. Breast Cancer Research and Treatment, 2018, 171, 719-735.	1.1	19
1450	Natural Selection Has Differentiated the Progesterone Receptor among Human Populations. American Journal of Human Genetics, 2018, 103, 45-57.	2.6	30
1451	Rare germline variants in DNA repair genes and the angiogenesis pathway predispose prostate cancer patients to develop metastatic disease. British Journal of Cancer, 2018, 119, 96-104.	2.9	40
1452	De novo ITPR1 variants are a recurrent cause of early-onset ataxia, acting via loss of channel function. European Journal of Human Genetics, 2018, 26, 1623-1634.	1.4	32
1453	Emerging applications of genome-editing technology to examine functionality of GWAS-associated variants for complex traits. Physiological Genomics, 2018, 50, 510-522.	1.0	17
1454	Abundance of clinical variants in exons included in multiple transcripts. Human Genomics, 2018, 12, 33.	1.4	2
1455	Molecular genetic study of Calpainopathy in Iran. Gene, 2018, 677, 259-265.	1.0	4

#	Article	IF	Citations
1456	The human CIB1â€“EVER1â€“EVER2 complex governs keratinocyte-intrinsic immunity to Î²-papillomaviruses. Journal of Experimental Medicine, 2018, 215, 2289-2310.	4.2	92
1457	Targeted next-generation sequencing as a comprehensive test for Mendelian diseases: a cohort diagnostic study. Scientific Reports, 2018, 8, 11646.	1.6	17
1458	Exome sequencing identifies novel ACE spliceâ€site variant in a fetus with renal tubular dysgenesis. Journal of Obstetrics and Gynaecology Research, 2018, 44, 2181-2185.	0.6	1
1459	Deciphering OPA1 mutations pathogenicity by combined analysis of human, mouse and yeast cell models. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 3496-3514.	1.8	36
1460	Whole-Exome Sequencing in Adults With Chronic Kidney Disease. Annals of Internal Medicine, 2018, 168, 100.	2.0	154
1461	An MTF1 binding site disrupted by a homozygous variant in the promoter of ATP7B likely causes Wilson Disease. European Journal of Human Genetics, 2018, 26, 1810-1818.	1.4	15
1462	Predicting the clinical impact of human mutation with deep neural networks. Nature Genetics, 2018, 50, 1161-1170.	9.4	288
1463	Arylsulphatase A Pseudodeficiency (ARSA-PD), hypertension and chronic renal disease in Aboriginal Australians. Scientific Reports, 2018, 8, 10912.	1.6	5
1464	Performance evaluation of pathogenicity-computation methods for missense variants. Nucleic Acids Research, 2018, 46, 7793-7804.	6.5	168
1465	Genetic association of the cytochrome c oxidase-related genes with Alzheimerâ€™s disease in Han Chinese. Neuropsychopharmacology, 2018, 43, 2264-2276.	2.8	29
1466	Somatic Mutations in Renal Cyst Epithelium in Autosomal Dominant Polycystic Kidney Disease. Journal of the American Society of Nephrology: JASN, 2018, 29, 2139-2156.	3.0	46
1467	Deep Scleral Exposure: A Degenerative Outcome of End-Stage Stargardt Disease. American Journal of Ophthalmology, 2018, 195, 16-25.	1.7	10
1468	Pancreatic cancer as a sentinel for hereditary cancer predisposition. BMC Cancer, 2018, 18, 697.	1.1	29
1469	Molecular Genetic Characterization of Patients With Focal Epilepsy Using a Customized Targeted Resequencing Gene Panel. Frontiers in Neurology, 2018, 9, 515.	1.1	16
1470	Identification of likely pathogenic and known variants in TSPEAR, LAMB3, BCOR, and WNT10A in four Turkish families with tooth agenesis. Human Genetics, 2018, 137, 689-703.	1.8	24
1471	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	2.6	69
1472	Actin cytoskeleton dynamics in stem cells from autistic individuals. Scientific Reports, 2018, 8, 11138.	1.6	29
1473	The Analysis of Variants in the General Population Reveals That PMM2 Is Extremely Tolerant to Missense Mutations and That Diagnosis of PMM2-CDG Can Benefit from the Identification of Modifiers. International Journal of Molecular Sciences, 2018, 19, 2218.	1.8	32

#	Article	IF	Citations
1474	Integrative Bioinformatics Approaches for Identification of Drug Targets in Hypertension. Frontiers in Cardiovascular Medicine, 2018, 5, 25.	1.1	3
1475	Next Generation Sequencing Methods for Diagnosis of Epilepsy Syndromes. Frontiers in Genetics, 2018, 9, 20.	1.1	102
1476	Alternative Splicing in Neurogenesis and Brain Development. Frontiers in Molecular Biosciences, 2018, 5, 12.	1.6	141
1477	Whole-Exome Sequencing in Searching for New Variants Associated With the Development of Parkinsonâ€™s Disease. Frontiers in Aging Neuroscience, 2018, 10, 136.	1.7	17
1478	Pleiotropic Effects of Variants in Dementia Genes in Parkinson Disease. Frontiers in Neuroscience, 2018, 12, 230.	1.4	21
1479	Exome Chip Analysis Identifies Low-Frequency and Rare Variants in MRPL38 for White Matter Hyperintensities on Brain Magnetic Resonance Imaging. Stroke, 2018, 49, 1812-1819.	1.0	17
1480	Heterozygous missense mutations in NFATC1 are associated with atrioventricular septal defect. Human Mutation, 2018, 39, 1428-1441.	1.1	15
1481	COL18A1 is a candidate eye iridocorneal angle-closure gene in humans. Human Molecular Genetics, 2018, 27, 3772-3786.	1.4	30
1482	Mutations in EMT-Related Genes in ALK Positive Crizotinib Resistant Non-Small Cell Lung Cancers. Cancers, 2018, 10, 10.	1.7	39
1483	IRF4 haploinsufficiency in a family with Whippleâ€™s disease. ELife, 2018, 7, .	2.8	43
1484	Bi-allelic Mutations in Phe-tRNA Synthetase Associated with a Multi-system Pulmonary Disease Support Non-translational Function. American Journal of Human Genetics, 2018, 103, 100-114.	2.6	34
1485	A Novel p.L145Q Mutation in the HNF1B Gene in a Case of Maturity-onset Diabetes of the Young Type 5 (MODY5). Internal Medicine, 2018, 57, 2035-2039.	0.3	6
1486	Lack of evidence for a causal role of CALR3 in monogenic cardiomyopathy. European Journal of Human Genetics, 2018, 26, 1603-1610.	1.4	4
1487	Landscape of pathogenic variations inÂa panel of 34 genes and cancer risk estimation from 5131 HBOC families. Genetics in Medicine, 2018, 20, 1677-1686.	1.1	62
1488	Cerebro-facio-thoracic dysplasia (Pascual-Castroviejo syndrome): Identification of a novel mutation, use of facial recognition analysis, and review of the literature. Translational Science of Rare Diseases, 2018, 3, 37-43.	1.6	5
1489	Phenotypic and genotypic overlap between mosaic NF2 and schwannomatosis in patients with multiple non-intradermal schwannomas. Human Genetics, 2018, 137, 543-552.	1.8	25
1490	Deep learning sequence-based ab initio prediction of variant effects on expression and disease risk. Nature Genetics, 2018, 50, 1171-1179.	9.4	375
1491	A <scp>PRRT</scp>2 variant in a Chinese family with paroxysmal kinesigenic dyskinesia and benign familial infantile seizures results in loss of interaction with <scp>STX</scp>1B. Epilepsia, 2018, 59, 1621-1630.	2.6	9

#	Article	IF	Citations
1492	Negative selection in tumor genome evolution acts on essential cellular functions and the immunopeptidome. Genome Biology, 2018, 19, 67.	3.8	81
1493	Exome sequencing of 85 Williamsâ€“Beuren syndrome cases rules out coding variation as a major contributor to remaining variance in social behavior. Molecular Genetics & Genomic Medicine, 2018, 6, 749-765.	0.6	9
1494	The conserved p.Arg108 residue in S1PR2 (DFNB68) is fundamental for proper hearing: evidence from a consanguineous Iranian family. BMC Medical Genetics, 2018, 19, 81.	2.1	10
1495	Prioritization and functional assessment of noncoding variants associated with complex diseases. Genome Medicine, 2018, 10, 53.	3.6	33
1496	Copy number variation analysis and targeted NGS in 77 families with suspected Lynch syndrome reveals novel potential causative genes. International Journal of Cancer, 2018, 143, 2800-2813.	2.3	11
1497	An Exome-wide Association Study for Type 2 Diabetesâ€“Attributed End-Stage Kidney Disease in African Americans. Kidney International Reports, 2018, 3, 867-878.	0.4	12
1498	Expanding Phenotype of Nephronophthisis-Related Ciliopathy: an Elderly Patient with Homozygous RPGRIP1L Mutation. Nephron, 2018, 140, 74-78.	0.9	2
1499	Whole exome sequencing reveals HSPA1L as a genetic risk factor for spontaneous preterm birth. PLoS Genetics, 2018, 14, e1007394.	1.5	35
1500	Letter to the Editor. Influence of rare RNF213 variants other than p.R4810K on the clinical outcomes of moyamoya disease. Journal of Neurosurgery, 2018, 129, 563-565.	0.9	5
1501	Molecular Deconvolution Platform to Establish Disease Mechanisms by Surveying GPCR Signaling. Cell Reports, 2018, 24, 557-568.e5.	2.9	12
1502	Analysis of causal effect of APOA5 variants on premature coronary artery disease. Annals of Human Genetics, 2018, 82, 437-447.	0.3	8
1503	A novel FBXO28 frameshift mutation in a child with developmental delay, dysmorphic features, and intractable epilepsy: A second gene that may contribute to the 1q41â€q42 deletion phenotype. American Journal of Medical Genetics, Part A, 2018, 176, 1549-1558.	0.7	9
1504	A De Novo POLD1 Mutation Associated With Mandibular Hypoplasia, Deafness, Progeroid Features, and Lipodystrophy Syndrome in a Family With Werner Syndrome. Journal of Investigative Medicine High Impact Case Reports, 2018, 6, 232470961878677.	0.3	10
1505	Longâ€range genomic regulators of THBS1 and LTBP4 modify disease severity in duchenne muscular dystrophy. Annals of Neurology, 2018, 84, 234-245.	2.8	53
1506	Identification of a missense variant in the WFS1 gene that causes a mild form of Wolfram syndrome and is associated with risk for type 2 diabetes in Ashkenazi Jewish individuals. Diabetologia, 2018, 61, 2180-2188.	2.9	38
1507	Rare variants in SOX17 are associated with pulmonary arterial hypertension with congenital heart disease. Genome Medicine, 2018, 10, 56.	3.6	112
1508	Evaluation of a bone morphogenetic protein 6 variant as a cause of iron loading. Human Genomics, 2018, 12, 23.	1.4	8
1510	Determination of the mutational landscape in Taiwanese patients with papillary thyroid cancer by whole-exome sequencing. Human Pathology, 2018, 78, 151-158.	1.1	9

#	Article	IF	Citations
1511	De novo mutations in MED13, a component of the Mediator complex, are associated with a novel neurodevelopmental disorder. Human Genetics, 2018, 137, 375-388.	1.8	46
1512	FANCM and RECQL genetic variants and breast cancer susceptibility: relevance to South Poland and West Ukraine. BMC Medical Genetics, 2018, 19, 12.	2.1	20
1513	Germline breast cancer susceptibility gene mutations and breast cancer outcomes. BMC Cancer, 2018, 18, 315.	1.1	61
1514	Deleterious genetic variants in ciliopathy genes increase risk of ritodrine-induced cardiac and pulmonary side effects. BMC Medical Genomics, 2018, 11, 4.	0.7	16
1515	Investigation of genetic variation and lifestyle determinants in vitamin D levels in Arab individuals. Journal of Translational Medicine, 2018, 16, 20.	1.8	9
1516	FineMAV: prioritizing candidate genetic variants driving local adaptations in human populations. Genome Biology, 2018, 19, 5.	3.8	20
1517	The VAAST Variant Prioritizer (VVP): ultrafast, easy to use whole genome variant prioritization tool. BMC Bioinformatics, 2018, 19, 57.	1.2	29
1518	Identification of missing variants by combining multiple analytic pipelines. BMC Bioinformatics, 2018, 19, 139.	1.2	10
1519	A Statistical Framework for Mapping Risk Genes from De Novo Mutations in Whole-Genome-Sequencing Studies. American Journal of Human Genetics, 2018, 102, 1031-1047.	2.6	26
1520	Preconception Carrier Screening by Genome Sequencing: Results from the Clinical Laboratory. American Journal of Human Genetics, 2018, 102, 1078-1089.	2.6	35
1521	Genetic Regulation of PCSK9 (Proprotein Convertase Subtilisin/Kexin Type 9) Plasma Levels and Its Impact on Atherosclerotic Vascular Disease Phenotypes. Circulation Genomic and Precision Medicine, 2018, 11, e001992.	1.6	37
1522	Partial loss-of-function of sodium channel SCN8A in familial isolated myoclonus. Human Mutation, 2018, 39, 965-969.	1.1	34
1523	Novel DCC variants in congenital mirror movements and evaluation of disease-associated missense variants. European Journal of Medical Genetics, 2018, 61, 329-334.	0.7	7
1524	Novel splice-site variant of UCHL1 in an Indian family with autosomal recessive spastic paraplegia-79. Journal of Human Genetics, 2018, 63, 927-933.	1.1	14
1525	Evidence for GALNT12 as a moderate penetrance gene for colorectal cancer. Human Mutation, 2018, 39, 1092-1101.	1.1	20
1526	Mutations affecting the actin regulator WD repeatâ€“containing protein 1 lead to aberrant lymphoid immunity. Journal of Allergy and Clinical Immunology, 2018, 142, 1589-1604.e11.	1.5	64
1527	Genomics of a pediatric ovarian fibrosarcoma. Association with the DICER1 syndrome. Scientific Reports, 2018, 8, 3252.	1.6	21
1528	Three-dimensional spatial analysis of missense variants in RTEL1 identifies pathogenic variants in patients with Familial Interstitial Pneumonia. BMC Bioinformatics, 2018, 19, 18.	1.2	18

#	Article	IF	Citations
1529	Lysosomal storage disorder gene variants in multiple system atrophy. Brain, 2018, 141, e53-e53.	3.7	11
1530	Functional variants in TBX2 are associated with a syndromic cardiovascular and skeletal developmental disorder. Human Molecular Genetics, 2018, 27, 2454-2465.	1.4	54
1531	INFERNO: inferring the molecular mechanisms of noncoding genetic variants. Nucleic Acids Research, 2018, 46, 8740-8753.	6.5	46
1532	Detection of Molecular Alterations in Taiwanese Patients with Medullary Thyroid Cancer Using Whole-Exome Sequencing. Endocrine Pathology, 2018, 29, 324-331.	5.2	23
1533	Analyzing the Effects of Genetic Variation in Noncoding Genomic Regions. , 2018, , 119-144.		3
1534	Modified penetrance of coding variants by cis-regulatory variation contributes to disease risk. Nature Genetics, 2018, 50, 1327-1334.	9.4	167
1535	Homozygous variants in KIAA1549, encoding a ciliary protein, are associated with autosomal recessive retinitis pigmentosa. Journal of Medical Genetics, 2018, 55, 705-712.	1.5	13
1536	High burden of birthweight-lowering genetic variants in Africans and Asians. BMC Medicine, 2018, 16, 70.	2.3	12
1537	Letter to the Editor. Multimodal opioid-minimizing pain management regimens following transsphenoidal surgery. Journal of Neurosurgery, 2018, 129, 561-562.	0.9	0
1538	Functional relevance for central cornea thickness-associated genetic variants by using integrative analyses. BioData Mining, 2018, 11, 19.	2.2	6
1539	Inherited CARD9 Deficiency: Invasive Disease Caused by Ascomycete Fungi in Previously Healthy Children and Adults. Journal of Clinical Immunology, 2018, 38, 656-693.	2.0	130
1540	First report of Klein-Waardenburg Syndrome in Iran and a novel pathogenic splice site variant in PAX3 gene. International Journal of Pediatric Otorhinolaryngology, 2018, 113, 229-233.	0.4	5
1541	Rare Variants in Known Susceptibility Loci and Their Contribution to Risk of Lung Cancer. Journal of Thoracic Oncology, 2018, 13, 1483-1495.	0.5	22
1542	Deep-coverage whole genome sequences and blood lipids among 16,324 individuals. Nature Communications, 2018, 9, 3391.	5.8	140
1543	A purely quantitative form of partial recessive IFN-Î³R2 deficiency caused by mutations of the initiation or second codon. Human Molecular Genetics, 2018, 27, 3919-3935.	1.4	14
1544	Bridging Genomics to Phenomics at Atomic Resolution through Variation Spatial Profiling. Cell Reports, 2018, 24, 2013-2028.e6.	2.9	33
1545	Life-threatening influenza pneumonitis in a child with inherited IRF9 deficiency. Journal of Experimental Medicine, 2018, 215, 2567-2585.	4.2	146
1546	Incomplete penetrance for isolated congenital asplenia in humans with mutations in translated and untranslated RPSA exons. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E8007-E8016.	3.3	31

#	Article	IF	Citations
1547	Elucidating the molecular pathogenesis of glioma: integrated germline and somatic profiling of a familial glioma case series. Neuro-Oncology, 2018, 20, 1625-1633.	0.6	12
1548	A comprehensive analysis of SNCAâ€related genetic risk in sporadic parkinson disease. Annals of Neurology, 2018, 84, 117-129.	2.8	50
1549	Identification of pathways and genes associated with cerebral palsy. Genes and Genomics, 2018, 40, 1339-1349.	0.5	8
1550	Realizing the significance of noncoding functionality in clinical genomics. Experimental and Molecular Medicine, 2018, 50, 1-8.	3.2	81
1551	Three VCP Mutations in Patients with Frontotemporal Dementia. Journal of Alzheimer's Disease, 2018, 65, 1139-1146.	1.2	19
1552	Genetic epidemiology of malignant hyperthermia in the UK. British Journal of Anaesthesia, 2018, 121, 944-952.	1.5	63
1553	Multiethnic meta-analysis identifies ancestry-specific and cross-ancestry loci for pulmonary function. Nature Communications, 2018, 9, 2976.	5.8	85
1554	Clonal dynamics of donor-derived myelodysplastic syndrome after unrelated hematopoietic cell transplantation for high-risk pediatric B-lymphoblastic leukemia. Journal of Physical Education and Sports Management, 2018, 4, a002980.	0.5	7
1555	Germline Variants and Risk for Pancreatic Cancer. Pancreas, 2018, 47, 924-936.	0.5	35
1556	The autism spectrum phenotype in ADNP syndrome. Autism Research, 2018, 11, 1300-1310.	2.1	49
1557	Expanding the spectrum of PEX16 mutations and novel insights into disease mechanisms. Molecular Genetics and Metabolism Reports, 2018, 16, 46-51.	0.4	21
1558	Familial Cancer Variant Prioritization Pipeline version 2 (FCVPPv2) applied to a papillary thyroid cancer family. Scientific Reports, 2018, 8, 11635.	1.6	30
1559	Targeted sequencing with expanded gene profile enables high diagnostic yield in non-5q-spinal muscular atrophies. Human Mutation, 2018, 39, 1284-1298.	1.1	42
1560	Genetic Analysis of Rare Human Variants of Regulators of G Protein Signaling Proteins and Their Role in Human Physiology and Disease. Pharmacological Reviews, 2018, 70, 446-474.	7.1	53
1561	Frequent and Rare HABP2 Variants Are Not Associated with Increased Susceptibility to Familial Nonmedullary Thyroid Carcinoma in the Spanish Population. Hormone Research in Paediatrics, 2018, 89, 397-407.	0.8	3
1562	Characterization of Heterozygous HTRA1 Mutations in Taiwanese Patients With Cerebral Small Vessel Disease. Stroke, 2018, 49, 1593-1601.	1.0	39
1563	Further delineation of Malan syndrome. Human Mutation, 2018, 39, 1226-1237.	1.1	42
1564	Comprehensive Cancer-Predisposition Gene Testing in an Adult Multiple Primary Tumor Series Shows a Broad Range of Deleterious Variants and Atypical Tumor Phenotypes. American Journal of Human Genetics, 2018, 103, 3-18.	2.6	46

#	Article	IF	Citations
1565	A recessive form of hyper-IgE syndrome by disruption of ZNF341-dependent STAT3 transcription and activity. Science Immunology, 2018, 3, .	5.6	132
1566	Estimating the mutational load for cardiovascular diseases in Pakistani population. PLoS ONE, 2018, 13, e0192446.	1.1	15
1567	Rare, potentially pathogenic variants in 21 keratoconus candidate genes are not enriched in cases in a large Australian cohort of European descent. PLoS ONE, 2018, 13, e0199178.	1.1	21
1568	Real-world clinical applicability of pathogenicity predictors assessed onSERPINA1mutations in alpha-1-antitrypsin deficiency. Human Mutation, 2018, 39, 1203-1213.	1.1	36
1569	A recurrent de novo missense mutation in UBTF causes developmental neuroregression. Human Molecular Genetics, 2018, 27, 691-705.	1.4	32
1570	Severe neurocognitive and growth disorders due to variation in THOC2 , an essential component of nuclear mRNA export machinery. Human Mutation, 2018, 39, 1126-1138.	1.1	28
1571	Assessing Rare Variation in Complex Traits. Methods in Molecular Biology, 2018, 1793, 51-71.	0.4	4
1572	An interactome perturbation framework prioritizes damaging missense mutations for developmental disorders. Nature Genetics, 2018, 50, 1032-1040.	9.4	64
1573	VarAFT: a variant annotation and filtration system for human next generation sequencing data. Nucleic Acids Research, 2018, 46, W545-W553.	6.5	136
1574	Role of Targeted Next Generation Sequencing in the Etiological Work-Up of Congenitally Deaf Children. Otology and Neurotology, 2018, 39, 732-738.	0.7	10
1575	Broad phenotypes in heterozygous NR5A1 46,XY patients with a disorder of sex development: an oligogenic origin?. European Journal of Human Genetics, 2018, 26, 1329-1338.	1.4	47
1576	Fine-mapping of prostate cancer susceptibility loci in a large meta-analysis identifies candidate causal variants. Nature Communications, 2018, 9, 2256.	5.8	88
1577	Alternative Splicing in Genetic Diseases: Improved Diagnosis and Novel Treatment Options. International Review of Cell and Molecular Biology, 2018, 335, 85-141.	1.6	23
1578	Novel mutations in the ALDH18A1 gene in complicated hereditary spastic paraplegia with cerebellar ataxia and cognitive impairment. Journal of Human Genetics, 2018, 63, 1009-1013.	1.1	18
1579	Genetic background of Japanese patients with pediatric hypertrophic and restrictive cardiomyopathy. Journal of Human Genetics, 2018, 63, 989-996.	1.1	26
1580	Germline Variation and Breast Cancer Incidence: A Gene-Based Association Study and Whole-Genome Prediction of Early-Onset Breast Cancer. Cancer Epidemiology Biomarkers and Prevention, 2018, 27, 1057-1064.	1.1	9
1581	A novel compound heterozygous mutation in the POMK gene causing limb-girdle muscular dystrophy-dystroglycanopathy in a sib pair. Neuromuscular Disorders, 2018, 28, 614-618.	0.3	11
1582	Recombination activity of human recombination-activating gene 2 (RAG2) mutations and correlation with clinical phenotype. Journal of Allergy and Clinical Immunology, 2019, 143, 726-735.	1.5	39

#	Article	IF	Citations
1583	Comprehensive annotation of BRCA1 and BRCA2 missense variants by functionally validated sequence-based computational prediction models. Genetics in Medicine, 2019, 21, 71-80.	1.1	52
1584	Evaluation of computational techniques for predicting non-synonymous single nucleotide variants pathogenicity. Genomics, 2019, 111, 869-882.	1.3	36
1585	Mouse models as a tool for discovering new neurological diseases. Neurobiology of Learning and Memory, 2019, 165, 106902.	1.0	17
1586	A novel EXT2 mutation in a consanguineous family with severe developmental delay, microcephaly, seizures, feeding difficulties, and osteopenia extends the phenotypic spectrum of autosomal recessive EXT2-related syndrome (AREXT2). European Journal of Medical Genetics, 2019, 62, 259-264.	0.7	11
1587	The Role of Next-Generation Sequencing in Pharmacogenetics and Pharmacogenomics. Cold Spring Harbor Perspectives in Medicine, 2019, 9, a033027.	2.9	49
1588	Implementing precision cancer medicine in the genomic era. Seminars in Cancer Biology, 2019, 55, 16-27.	4.3	24
1589	ncdDetect2: improved models of the site-specific mutation rate in cancer and driver detection with robust significance evaluation. Bioinformatics, 2019, 35, 189-199.	1.8	6
1590	Molecular Methods. , 2019, , 1297-1310.e1.		0
1591	Regulatory variants: from detection to predicting impact. Briefings in Bioinformatics, 2019, 20, 1639-1654.	3.2	82
1592	Seave: a comprehensive web platform for storing and interrogating human genomic variation. Bioinformatics, 2019, 35, 122-125.	1.8	26
1593	Predicting Non-Synonymous Single Nucleotide Variants Pathogenic Effects in Human Diseases. , 2019, , 400-409.		1
1594	Genetic and Transcriptomic Variation Linked to Neutrophil Granulocyteâ€“Macrophage Colony-Stimulating Factor Signaling in Pediatric Crohnâ€™s Disease. Inflammatory Bowel Diseases, 2019, 25, 547-560.	0.9	8
1595	Deleterious alleles in the context of domestication, inbreeding, and selection. Evolutionary Applications, 2019, 12, 6-17.	1.5	94
1596	Novel DLX3 variants in amelogenesis imperfecta with attenuated trichoâ€dentoâ€osseous syndrome. Oral Diseases, 2019, 25, 182-191.	1.5	17
1597	Computational resources associating diseases with genotypes, phenotypes and exposures. Briefings in Bioinformatics, 2019, 20, 2098-2115.	3.2	27
1598	Detecting and Annotating Rare Variants. , 2019, , 388-399.		4
1599	Major intra-familial phenotypic heterogeneity and incomplete penetrance due to a CACNA1A pathogenic variant. European Journal of Medical Genetics, 2019, 62, 103530.	0.7	34
1600	Noncardiac genetic predisposition in sudden infant death syndrome. Genetics in Medicine, 2019, 21, 641-649.	1.1	9

#	Article	IF	Citations
1601	Bi-allelic c.181_183delTGT in BTB domain of KLHL7 is associated with overlapping phenotypes of Crisponi/CISS1-like and Bohring-Opitz like syndrome. European Journal of Medical Genetics, 2019, 62, 103528.	0.7	7
1602	Fine-mapping analysis of a chromosome 2 region linked to resistance to Mycobacterium tuberculosis infection in Uganda reveals potential regulatory variants. Genes and Immunity, 2019, 20, 473-483.	2.2	18
1603	Clinical and genetic differences between pustular psoriasis subtypes. Journal of Allergy and Clinical Immunology, 2019, 143, 1021-1026.	1.5	165
1604	Genome-wide meta-analysis of macronutrient intake of 91,114 European ancestry participants from the cohorts for heart and aging research in genomic epidemiology consortium. Molecular Psychiatry, 2019, 24, 1920-1932.	4.1	44
1605	Health behaviors among unaffected participants following receipt of variants of uncertain significance in cardiomyopathy-associated genes. Genetics in Medicine, 2019, 21, 748-752.	1.1	3
1606	Phrank measures phenotype sets similarity to greatly improve Mendelian diagnostic disease prioritization. Genetics in Medicine, 2019, 21, 464-470.	1.1	33
1607	dbCID: a manually curated resource for exploring the driver indels in human cancer. Briefings in Bioinformatics, 2019, 20, 1925-1933.	3.2	11
1608	A Bioinformatics Toolkit: In Silico Tools and Online Resources for Investigating Genetic Variation. Seminars in Thrombosis and Hemostasis, 2019, 45, 674-684.	1.5	1
1609	Predicting the Occurrence of Variants in RAG1 and RAG2. Journal of Clinical Immunology, 2019, 39, 688-701.	2.0	3
1610	Prioritizing natural-selection signals from the deep-sequencing genomic data suggests multi-variant adaptation in Tibetan highlanders. National Science Review, 2019, 6, 1201-1222.	4.6	30
1611	Family-Based Quantitative Trait Meta-Analysis Implicates Rare Noncoding Variants in DENND1A in Polycystic Ovary Syndrome. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 3835-3850.	1.8	51
1612	Mutations in PLS1 , encoding fimbrin, cause autosomal dominant nonsyndromic hearing loss. Human Mutation, 2019, 40, 2286-2295.	1.1	23
1613	Saturation mutagenesis of twenty disease-associated regulatory elements at single base-pair resolution. Nature Communications, 2019, 10, 3583.	5.8	152
1614	Reevaluation of genetic variants previously associated with arrhythmogenic right ventricular cardiomyopathy integrating populationâ€based cohorts and proteomics data. Clinical Genetics, 2019, 96, 506-514.	1.0	14
1615	Direct Evidence of an Increasing Mutational Load in Humans. Molecular Biology and Evolution, 2019, 36, 2823-2829.	3.5	12
1616	Multi-Modality Analysis Improves Survival Prediction in Enucleated Uveal Melanoma Patients. , 2019, 60, 3595.		12
1617	Genetic variants of HIF1Î± are associated with right ventricular fibrotic load in repaired tetralogy of Fallot patients: a cardiovascular magnetic resonance study. Journal of Cardiovascular Magnetic Resonance, 2019, 21, 51.	1.6	5
1618	ALG9 Mutation Carriers Develop Kidney and Liver Cysts. Journal of the American Society of Nephrology: JASN, 2019, 30, 2091-2102.	3.0	91

#	Article	IF	Citations
1619	Bi-allelic GOT2 Mutations Cause a Treatable Malate-Aspartate Shuttle-Related Encephalopathy. American Journal of Human Genetics, 2019, 105, 534-548.	2.6	46
1620	Beyond the Exome: The Non-coding Genome and Enhancers in Neurodevelopmental Disorders and Malformations of Cortical Development. Frontiers in Cellular Neuroscience, 2019, 13, 352.	1.8	53
1621	Biallelic and De Novo Variants in DONSON Reveal a Clinical Spectrum of Cell Cycleâ€opathies with Microcephaly, Dwarfism and Skeletal Abnormalities. American Journal of Medical Genetics, Part A, 2019, 179, 2056-2066.	0.7	15
1622	MAGEL2â€related disorders: A study and case series. Clinical Genetics, 2019, 96, 493-505.	1.0	26
1623	Whole genome sequencing and rare variant analysis in essential tremor families. PLoS ONE, 2019, 14, e0220512.	1.1	28
1624	A de novo variant in the Xâ€linked gene CNKSR2 is associated with seizures and mild intellectual disability in a female patient. Molecular Genetics & Genomic Medicine, 2019, 7, e00861.	0.6	13
1625	Sequencing reveals protective and pathogenic effects on development of diabetes of rare GLIS3 variants. PLoS ONE, 2019, 14, e0220805.	1.1	4
1626	Challenges in funding and developing genomic software: roots and remedies. Genome Biology, 2019, 20, 147.	3.8	21
1628	Identifying Putative Susceptibility Genes and Evaluating Their Associations with Somatic Mutations in Human Cancers. American Journal of Human Genetics, 2019, 105, 477-492.	2.6	27
1629	Rare De Novo Missense Variants in RNA Helicase DDX6 Cause Intellectual Disability and Dysmorphic Features and Lead to P-Body Defects and RNA Dysregulation. American Journal of Human Genetics, 2019, 105, 509-525.	2.6	50
1630	A regression framework to uncover pleiotropy in large-scale electronic health record data. Journal of the American Medical Informatics Association: JAMIA, 2019, 26, 1083-1090.	2.2	9
1631	Rare and common variant discovery in complex disease: the IBD case study. Human Molecular Genetics, 2019, 28, R162-R169.	1.4	13
1632	Contributions of Rare Gene Variants to Familial and Sporadic FSGS. Journal of the American Society of Nephrology: JASN, 2019, 30, 1625-1640.	3.0	42
1633	Using game theory and decision decomposition to effectively discern and characterise bi-locus diseases. Artificial Intelligence in Medicine, 2019, 99, 101690.	3.8	10
1634	A Novel Mutation in MARS in a Patient with Charcot-Marie-Tooth Disease, Axonal, Type 2U with Congenital Onset. Journal of Neuromuscular Diseases, 2019, 6, 333-339.	1.1	12
1635	Next generation sequencing study in a cohort of Italian patients with syndromic hearing loss. Hearing Research, 2019, 381, 107769.	0.9	7
1636	Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes. Npj Genomic Medicine, 2019, 4, 19.	1.7	163
1637	Human DEF6 deficiency underlies an immunodeficiency syndrome with systemic autoimmunity and aberrant CTLA-4 homeostasis. Nature Communications, 2019, 10, 3106.	5.8	48

#	Article	IF	Citations
1638	Benchmarking subcellular localization and variant tolerance predictors on membrane proteins. BMC Genomics, 2019, 20, 547.	1.2	14
1639	Assessing predictions of the impact of variants on splicing in CAGI5. Human Mutation, 2019, 40, 1215-1224.	1.1	18
1640	Statistical representation models for mutation information within genomic data. BMC Bioinformatics, 2019, 20, 324.	1.2	4
1641	Gene-based genome-wide association studies and meta-analyses of conotruncal heart defects. PLoS ONE, 2019, 14, e0219926.	1.1	15
1642	A novel WARS mutation causes distal hereditary motor neuropathy in a Chinese family. Brain, 2019, 142, e49-e49.	3.7	12
1643	CAGI5: Objective performance assessments of predictions based on the Evolutionary Action equation. Human Mutation, 2019, 40, 1436-1454.	1.1	26
1644	Genome-wide analysis indicates association between heterozygote advantage and healthy aging in humans. BMC Genetics, 2019, 20, 52.	2.7	10
1645	Sudden unexpected cardiac death and postmortem identification of a novel RYR2 gene mutation. International Journal of Legal Medicine, 2019, 133, 1835-1838.	1.2	3
1646	WITER: a powerful method for estimation of cancer-driver genes using a weighted iterative regression modelling background mutation counts. Nucleic Acids Research, 2019, 47, e96-e96.	6.5	28
1647	GATA6 mutations: Characterization of two novel patients and a comprehensive overview of the GATA6 genotypic and phenotypic spectrum. American Journal of Medical Genetics, Part A, 2019, 179, 1836-1845.	0.7	16
1648	Identifying Cancer Driver lncRNAs Bridged by Functional Effectors through Integrating Multi-omics Data in Human Cancers. Molecular Therapy - Nucleic Acids, 2019, 17, 362-373.	2.3	30
1649	Biallelic variants in CTU2 cause DREAMâ€PL syndrome and impair thiolation of tRNA wobble U34. Human Mutation, 2019, 40, 2108-2120.	1.1	25
1650	Gene therapy for primary immunodeficiency. Human Molecular Genetics, 2019, 28, R15-R23.	1.4	55
1651	Building a Hybrid Physical-Statistical Classifier for Predicting the Effect of Variants Related to Protein-Drug Interactions. Structure, 2019, 27, 1469-1481.e3.	1.6	6
1652	De novo substitutions of TRPM3 cause intellectual disability and epilepsy. European Journal of Human Genetics, 2019, 27, 1611-1618.	1.4	45
1653	Systematic discovery of conservation states for single-nucleotide annotation of the human genome. Communications Biology, 2019, 2, 248.	2.0	15
1654	Infantile-Onset Paroxysmal Movement Disorder and Episodic Ataxia Associated with a TBC1D24 Mutation. Neuropediatrics, 2019, 50, 308-312.	0.3	12
1655	VIPdb, a genetic Variant Impact Predictor Database. Human Mutation, 2019, 40, 1202-1214.	1.1	24

#	Article	IF	Citations
1656	Adopting High-Resolution Allele Frequencies Substantially Expedites Variant Interpretation in Genetic Diagnostic Laboratories. Journal of Molecular Diagnostics, 2019, 21, 602-611.	1.2	0
1657	Systematic Review of Somatic Mutations in Splenic Marginal Zone Lymphoma. Scientific Reports, 2019, 9, 10444.	1.6	23
1658	GemC1 is a critical switch for neural stem cell generation in the postnatal brain. Glia, 2019, 67, 2360-2373.	2.5	23
1659	Exome sequencing of family trios from the National Birth Defects Prevention Study: Tapping into a rich resource of genetic and environmental data. Birth Defects Research, 2019, 111, 1618-1632.	0.8	9
1660	Genetic association testing using the GENESIS R/Bioconductor package. Bioinformatics, 2019, 35, 5346-5348.	1.8	260
1661	Cohesin complex-associated holoprosencephaly. Brain, 2019, 142, 2631-2643.	3.7	43
1662	Addressing the Missing Heritability Problem With the Help of Regulatory Features. Evolutionary Bioinformatics, 2019, 15, 117693431986086.	0.6	0
1663	A Deep Belief Networks Based Prediction Method for Identification of Disease-Associated Non-coding SNPs in Human Genome. Lecture Notes in Computer Science, 2019, , 12-24.	1.0	0
1664	A brain somatic RHEB doublet mutation causes focal cortical dysplasia type II. Experimental and Molecular Medicine, 2019, 51, 1-11.	3.2	46
1665	Quantifying gene selection in cancer through protein functional alteration bias. Nucleic Acids Research, 2019, 47, 6642-6655.	6.5	21
1666	De Novo Heterozygous POLR2A Variants Cause a Neurodevelopmental Syndrome with Profound Infantile-Onset Hypotonia. American Journal of Human Genetics, 2019, 105, 283-301.	2.6	46
1667	Actionable Pharmacogenetic Variation in the Slovenian Genomic Database. Frontiers in Pharmacology, 2019, 10, 240.	1.6	10
1668	TASP1 mutation in a female with craniofacial anomalies, anterior segment dysgenesis, congenital immunodeficiency and macrocytic anemia. Molecular Genetics & Genomic Medicine, 2019, 7, e818.	0.6	6
1669	Functional Characterization of Rare Variants in the SHOX2 Gene Identified in Sinus Node Dysfunction and Atrial Fibrillation. Frontiers in Genetics, 2019, 10, 648.	1.1	21
1670	Host Genetics, Innate Immune Responses, and Cellular Death Pathways in Poliomyelitis Patients. Frontiers in Microbiology, 2019, 10, 1495.	1.5	7
1671	Estimation of allele-specific fitness effects across human protein-coding sequences and implications for disease. Genome Research, 2019, 29, 1310-1321.	2.4	24
1672	Rare DEGS1 variant significantly alters de novo ceramide synthesis pathway. Journal of Lipid Research, 2019, 60, 1630-1639.	2.0	16
1673	Family with primary periodic paralysis and a mutation in MCM3AP, a gene implicated in mRNA transport. Muscle and Nerve, 2019, 60, 311-314.	1.0	3

#	Article	IF	Citations
1674	NovelACTN1variants in cases of thrombocytopenia. Human Mutation, 2019, 40, 2258-2269.	1.1	5
1675	Diagnostic Yield and Treatment Impact of Targeted Exome Sequencing in Early-Onset Epilepsy. Frontiers in Neurology, 2019, 10, 434.	1.1	70
1676	Predicting changes in protein stability caused by mutation using sequenceâ€and structureâ€based methods in a CAGI5 blind challenge. Human Mutation, 2019, 40, 1414-1423.	1.1	28
1677	Genetic Variation, Comparative Genomics, and the Diagnosis of Disease. New England Journal of Medicine, 2019, 381, 64-74.	13.9	127
1678	Inherited IFNAR1 deficiency in otherwise healthy patients with adverse reaction to measles and yellow fever live vaccines. Journal of Experimental Medicine, 2019, 216, 2057-2070.	4.2	127
1679	De Novo Variants in TAOK1 Cause Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 105, 213-220.	2.6	36
1680	Paralog Studies Augment Gene Discovery: DDX and DHX Genes. American Journal of Human Genetics, 2019, 105, 302-316.	2.6	56
1681	Common BACE2 Polymorphisms are Associated with Altered Risk for Alzheimerâ€™s Disease and CSF Amyloid Biomarkers in APOE Îµ4 Non-Carriers. Scientific Reports, 2019, 9, 9640.	1.6	14
1682	Genetic diagnosis in first or second trimester pregnancy loss using exome sequencing: a systematic review of human essential genes. Journal of Assisted Reproduction and Genetics, 2019, 36, 1539-1548.	1.2	25
1683	RegulationSpotter: annotation and interpretation of extratranscriptic DNA variants. Nucleic Acids Research, 2019, 47, W106-W113.	6.5	17
1684	Bayesian modeling to predict malignant hyperthermia susceptibility and pathogenicity of RYR1, CACNA1S and STAC3 variants. Pharmacogenomics, 2019, 20, 989-1003.	0.6	1
1685	Coexistence of Congenital Adrenal Hyperplasia and Autoimmune Addison's Disease. Frontiers in Endocrinology, 2019, 10, 648.	1.5	2
1686	Spectrum of K_V2.1 Dysfunction in KCNB1â€Associated Neurodevelopmental Disorders. Annals of Neurology, 2019, 86, 899-912.	2.8	52
1687	SETD2 related overgrowth syndrome: Presentation of four new patients and review of the literature. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 509-518.	0.7	24
1688	Towards genomic database of Alexander disease to identify variations modifying disease phenotype. Scientific Reports, 2019, 9, 14763.	1.6	10
1689	Whole Genome Sequencing of Familial Non-Medullary Thyroid Cancer Identifies Germline Alterations in MAPK/ERK and PI3K/AKT Signaling Pathways. Biomolecules, 2019, 9, 605.	1.8	27
1690	Essentials of Bioinformatics, Volume II. , 2019, , .		1
1691	Pathogenicity discrimination and genetic test reference for CRX variants based on genotype-phenotype analysis. Experimental Eye Research, 2019, 189, 107846.	1.2	18

#	Article	IF	Citations
1692	Mutation analysis of LRP10 in Japanese patients with familial Parkinson's disease, progressive supranuclear palsy, and frontotemporal dementia. Neurobiology of Aging, 2019, 84, 235.e11-235.e16.	1.5	10
1693	Bardet-Biedl Syndrome in rhesus macaques: A nonhuman primate model of retinitis pigmentosa. Experimental Eye Research, 2019, 189, 107825.	1.2	35
1694	Rare-variant collapsing analyses for complex traits: guidelines and applications. Nature Reviews Genetics, 2019, 20, 747-759.	7.7	147
1695	Evidence of Neutral Evolution of Mitochondrial DNA in Human Hepatocellular Carcinoma. Genome Biology and Evolution, 2019, 11, 2909-2916.	1.1	3
1696	A novel disorder involving dyshematopoiesis, inflammation, and HLH due to aberrant CDC42 function. Journal of Experimental Medicine, 2019, 216, 2778-2799.	4.2	132
1697	Informing disease modelling with brain-relevant functional genomic annotations. Brain, 2019, 142, 3694-3712.	3.7	8
1698	Toward Clinical Implementation of Next-Generation Sequencing-Based Genetic Testing in Rare Diseases: Where Are We?. Trends in Genetics, 2019, 35, 852-867.	2.9	65
1699	Rare mutations in the complement regulatory gene CSMD1 are associated with male and female infertility. Nature Communications, 2019, 10, 4626.	5.8	24
1700	Circulating Oxytocin Is Genetically Determined and Associated With Obesity and Impaired Glucose Tolerance. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 5621-5632.	1.8	14
1701	IFT172 as the 19th gene causative of oralâ€facialâ€digital syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 2510-2513.	0.7	9
1702	Metaâ€MultiSKAT: Multiple phenotype metaâ€analysis for regionâ€based association test. Genetic Epidemiology, 2019, 43, 800-814.	0.6	9
1703	De novo pathogenic DNM1L variant in a patient diagnosed with atypical hereditary sensory and autonomic neuropathy. Molecular Genetics & Genomic Medicine, 2019, 7, e00961.	0.6	12
1704	Gene4Denovo: an integrated database and analytic platform for de novo mutations in humans. Nucleic Acids Research, 2020, 48, D913-D926.	6.5	41
1705	Single-step access to a series of Dâ€“A Ï€-conjugated oligomers with 3â€“10 nm chain lengths. Polymer Chemistry, 2019, 10, 325-330.	1.9	15
1706	De novo Mutations From Whole Exome Sequencing in Neurodevelopmental and Psychiatric Disorders: From Discovery to Application. Frontiers in Genetics, 2019, 10, 258.	1.1	49
1707	Phenome-wide association analysis of LDL-cholesterol lowering genetic variants in PCSK9. BMC Cardiovascular Disorders, 2019, 19, 240.	0.7	22
1708	A de novo SCN8A heterozygous mutation in a child with epileptic encephalopathy: a case report. BMC Pediatrics, 2019, 19, 400.	0.7	3
1709	First genome-wide association study of non-severe malaria in two birth cohorts in Benin. Human Genetics, 2019, 138, 1341-1357.	1.8	14

#	Article	IF	Citations
1710	Germline variants associated with leukocyte genes predict tumor recurrence in breast cancer patients. Npj Precision Oncology, 2019, 3, 28.	2.3	19
1711	A-to-I RNA editing contributes to the persistence of predicted damaging mutations in populations. Genome Research, 2019, 29, 1766-1776.	2.4	5
1712	VariCarta: A Comprehensive Database of Harmonized Genomic Variants Found in Autism Spectrum Disorder Sequencing Studies. Autism Research, 2019, 12, 1728-1736.	2.1	23
1713	Genotypeâ€“Phenotype Association Analysis Reveals New Pathogenic Factors for Osteogenesis Imperfecta Disease. Frontiers in Pharmacology, 2019, 10, 1200.	1.6	11
1714	The genetic landscape of the human solute carrier (SLC) transporter superfamily. Human Genetics, 2019, 138, 1359-1377.	1.8	79
1715	Disease modeling of a mutation in Î±â€actinin 2 guides clinical therapy in hypertrophic cardiomyopathy. EMBO Molecular Medicine, 2019, 11, e11115.	3.3	88
1716	Linkage and exome analysis implicate multiple genes in non-syndromic intellectual disability in a large Swedish family. BMC Medical Genomics, 2019, 12, 156.	0.7	3
1717	De novo NSF mutations cause early infantile epileptic encephalopathy. Annals of Clinical and Translational Neurology, 2019, 6, 2334-2339.	1.7	10
1718	Genetic variability in Iranian limbâ€girdle muscular dystrophy type 2B patients: An evidence of a founder effect. Molecular Genetics & Genomic Medicine, 2019, 7, e1029.	0.6	2
1719	CAUSALdb: a database for disease/trait causal variants identified using summary statistics of genome-wide association studies. Nucleic Acids Research, 2019, 48, D807-D816.	6.5	34
1720	Rare singleâ€nucleotide variants in oculoâ€auriculoâ€vertebral spectrum (OAVS). Molecular Genetics & Genomic Medicine, 2019, 7, e00959.	0.6	10
1721	Predicting Functional Effects of Synonymous Variants: A Systematic Review and Perspectives. Frontiers in Genetics, 2019, 10, 914.	1.1	67
1722	Sequencing Analysis at 8p23 Identifies Multiple Rare Variants in DLC1 Associated with Sleep-Related Oxyhemoglobin Saturation Level. American Journal of Human Genetics, 2019, 105, 1057-1068.	2.6	10
1723	Identification and functional characterization of mutations within HADHB associated with mitochondrial trifunctional protein deficiency. Mitochondrion, 2019, 49, 200-205.	1.6	6
1724	Relating Aerial Deposition of Entomophaga maimaiga Conidia (Zoopagomycota: Entomophthorales) to Mortality of Gypsy Moth (Lepidoptera: Erebidae) Larvae and Nearby Defoliation. Environmental Entomology, 2019, 48, 1214-1222.	0.7	13
1725	The Genetic Architecture of Parkinson Disease in Spain: Characterizing Populationâ€Specific Risk, Differential Haplotype Structures, and Providing Etiologic Insight. Movement Disorders, 2019, 34, 1851-1863.	2.2	47
1726	Chromosomal microarray and wholeâ€exome sequence analysis in Taiwanese patients with autism spectrum disorder. Molecular Genetics & Genomic Medicine, 2019, 7, e996.	0.6	11
1727	RPL13 Variants Cause Spondyloepimetaphyseal Dysplasia with Severe Short Stature. American Journal of Human Genetics, 2019, 105, 1040-1047.	2.6	17

#	Article	IF	Citations
1728	PGG.SNV: understanding the evolutionary and medical implications of human single nucleotide variations in diverse populations. Genome Biology, 2019, 20, 215.	3.8	30
1729	SLC12A ion transporter mutations in sporadic and familial human congenital hydrocephalus. Molecular Genetics & Genomic Medicine, 2019, 7, e892.	0.6	22
1730	A unified encyclopedia of human functional DNA elements through fully automated annotation of 164 human cell types. Genome Biology, 2019, 20, 180.	3.8	37
1731	MAPT p.V363I mutation. Neurology: Genetics, 2019, 5, e347.	0.9	10
1732	Targeted sequencing of linkage region in Dominican families implicates PRIMA1 and the SPATA7-PTPN21-ZC3H14-EML5-TTC8 locus in carotid-intima media thickness and atherosclerotic events. Scientific Reports, 2019, 9, 11621.	1.6	0
1733	Targeted sequencing of circulating cell-free DNA in stage II-III resectable oesophageal squamous cell carcinoma patients. BMC Cancer, 2019, 19, 818.	1.1	16
1734	A region-based gene association study combined with a leave-one-out sensitivity analysis identifies SMG1 as a pancreatic cancer susceptibility gene. PLoS Genetics, 2019, 15, e1008344.	1.5	13
1735	Association of Germline Variants in Natural Killer Cells With Tumor Immune Microenvironment Subtypes, Tumor-Infiltrating Lymphocytes, Immunotherapy Response, Clinical Outcomes, and Cancer Risk. JAMA Network Open, 2019, 2, e199292.	2.8	49
1736	GRAM: A GeneRAlized Model to predict the molecular effect of a non-coding variant in a cell-type specific manner. PLoS Genetics, 2019, 15, e1007860.	1.5	1
1737	Genome-wide association analysis of 95â€‰549 individuals identifies novel loci and genes influencing optic disc morphology. Human Molecular Genetics, 2019, 28, 3680-3690.	1.4	19
1738	The formation of human populations in South and Central Asia. Science, 2019, 365, .	6.0	383
1739	A Novel Homozygous Deletion within the <i>FRY</i> Gene Associated with Nonsyndromic Developmental Delay. Cytogenetic and Genome Research, 2019, 159, 19-25.	0.6	5
1740	Low-frequency variants at the CYP2C9 locus among Puerto Rican patients on warfarin: in silico predictions of functionality and conservation. Pharmacogenomics, 2019, 20, 891-902.	0.6	2
1741	de novo MEPCE nonsense variant associated with a neurodevelopmental disorder causes disintegration of 7SK snRNP and enhanced RNA polymerase II activation. Scientific Reports, 2019, 9, 12516.	1.6	13
1742	Redefining the Etiologic Landscape of Cerebellar Malformations. American Journal of Human Genetics, 2019, 105, 606-615.	2.6	61
1743	Screening of 31 genes involved in monogenic forms of obesity in 23 Pakistani probands with early-onset childhood obesity: a case report. BMC Medical Genetics, 2019, 20, 152.	2.1	3
1744	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogryposis. American Journal of Human Genetics, 2019, 105, 689-705.	2.6	48
1745	REVEL and BayesDel outperform other in silico meta-predictors for clinical variant classification. Scientific Reports, 2019, 9, 12752.	1.6	52

#	Article	IF	Citations
1746	The Evolving Field of Genetic Epidemiology: From Familial Aggregation to Genomic Sequencing. American Journal of Epidemiology, 2019, 188, 2069-2077.	1.6	6
1747	regBase: whole genome base-wise aggregation and functional prediction for human non-coding regulatory variants. Nucleic Acids Research, 2019, 47, e134-e134.	6.5	41
1748	Genome-wide association meta-analyses and fine-mapping elucidate pathways influencing albuminuria. Nature Communications, 2019, 10, 4130.	5.8	133
1749	Exome Sequencing Reveals Immune Genes as Susceptibility Modifiers in Individuals with Î±1-Antitrypsin Deficiency. Scientific Reports, 2019, 9, 13088.	1.6	7
1750	Rare variants in FANCA induce premature ovarian insufficiency. Human Genetics, 2019, 138, 1227-1236.	1.8	56
1751	Targeted next-generation DNA sequencing identifies Notch signaling pathway mutation as a predictor of radiation response. International Journal of Radiation Biology, 2019, 95, 1640-1647.	1.0	2
1752	Druggable Transcriptional Networks in the Human Neurogenic Epigenome. Pharmacological Reviews, 2019, 71, 520-538.	7.1	11
1753	A missense variant in PER2 is associated with delayed sleepâ€“wake phase disorder in a Japanese population. Journal of Human Genetics, 2019, 64, 1219-1225.	1.1	19
1754	Mid-Frequency Hearing Loss Is Characteristic Clinical Feature of OTOA-Associated Hearing Loss. Genes, 2019, 10, 715.	1.0	15
1755	De novo variants in PAK1 lead to intellectual disability with macrocephaly and seizures. Brain, 2019, 142, 3351-3359.	3.7	29
1756	Heteroplasmy concordance between mitochondrial DNA and RNA. Scientific Reports, 2019, 9, 12942.	1.6	9
1757	A small gene sequencing panel realises a high diagnostic rate in patients with congenital nystagmus following basic phenotyping. Scientific Reports, 2019, 9, 13229.	1.6	9
1758	NLRX1 inhibits the early stages of CNS inflammation and prevents the onset of spontaneous autoimmunity. PLoS Biology, 2019, 17, e3000451.	2.6	21
1759	Blau syndrome with a rare mutation in exon 9 of NOD2 gene. Autoimmunity, 2019, 52, 256-263.	1.2	3
1760	Postzygotic inactivating mutations of RHOA cause a mosaic neuroectodermal syndrome. Nature Genetics, 2019, 51, 1438-1441.	9.4	25
1761	Bridging the Gaps in Personalized Medicine Value Assessment: A Review of the Need for Outcome Metrics across Stakeholders and Scientific Disciplines. Public Health Genomics, 2019, 22, 16-24.	0.6	9
1762	The Prevalence and Clinical Characteristics of TECTA-Associated Autosomal Dominant Hearing Loss. Genes, 2019, 10, 744.	1.0	17
1763	Pathway analysis of genomic pathology tests for prognostic cancer subtyping. Journal of Biomedical Informatics, 2019, 98, 103286.	2.5	3

#	Article	IF	Citations
1764	Target genes, variants, tissues and transcriptional pathways influencing human serum urate levels. Nature Genetics, 2019, 51, 1459-1474.	9.4	251
1765	Detailed Clinical Features of Deafness Caused by a Claudin-14 Variant. International Journal of Molecular Sciences, 2019, 20, 4579.	1.8	4
1766	Ancestry-Dependent Enrichment of Deleterious Homozygotes in Runs of Homozygosity. American Journal of Human Genetics, 2019, 105, 747-762.	2.6	36
1767	Estimating the Frequency of Single Point Driver Mutations across Common Solid Tumours. Scientific Reports, 2019, 9, 13452.	1.6	6
1768	Broad Phenotypes of Disorders/Differences of Sex Development in MAMLD1 Patients Through Oligogenic Disease. Frontiers in Genetics, 2019, 10, 746.	1.1	22
1769	Genome-wide association study on antipsychotic-induced weight gain in Europeans and African-Americans. Schizophrenia Research, 2019, 212, 204-212.	1.1	15
1770	Genetic and epigenetic study of an Alzheimerâ€™s disease family with monozygotic triplets. Brain, 2019, 142, 3375-3381.	3.7	11
1771	Unusual Segregation of APP Mutations in Monogenic Alzheimer Disease. Neurodegenerative Diseases, 2019, 19, 96-100.	0.8	3
1772	Introduction to Human Genetics. , 2019, , 1-17.		1
1773	Modification of the PROM1 disease phenotype by a mutation in ABCA4. Ophthalmic Genetics, 2019, 40, 369-375.	0.5	17
1774	Functional interpretation of genetic variants using deep learning predicts impact on chromatin accessibility and histone modification. Nucleic Acids Research, 2019, 47, 10597-10611.	6.5	39
1775	Identification of Polycystic Kidney Disease 1 Like 1 Gene Variants in Children With Biliary Atresia Splenic Malformation Syndrome. Hepatology, 2019, 70, 899-910.	3.6	58
1776	Biological Interpretation of Complex Genomic Data. Methods in Molecular Biology, 2019, 1908, 61-71.	0.4	1
1777	A significant inflation in TGM6 genetic risk casts doubt in its causation in spinocerebellar ataxia type 35. Parkinsonism and Related Disorders, 2019, 63, 42-45.	1.1	11
1778	Novel genetic and epigenetic factors of importance for inter-individual differences in drug disposition, response and toxicity. , 2019, 197, 122-152.		83
1779	Low-frequency variation in TP53 has large effects on head circumference and intracranial volume. Nature Communications, 2019, 10, 357.	5.8	30
1780	Xrare: a machine learning method jointly modeling phenotypes and genetic evidence for rare disease diagnosis. Genetics in Medicine, 2019, 21, 2126-2134.	1.1	56
1781	The prevalence of germline DICER1 pathogenic variation in cancer populations. Molecular Genetics & Genomic Medicine, 2019, 7, e555.	0.6	24

#	Article	IF	Citations
1782	Contribution of rare and common variants to intellectual disability in a sub-isolate of Northern Finland. Nature Communications, 2019, 10, 410.	5.8	32
1783	A suite of automated sequence analyses reduces the number of candidate deleterious variants and reveals a difference between probands and unaffected siblings. Genetics in Medicine, 2019, 21, 1772-1780.	1.1	7
1784	GenESysV: a fast, intuitive and scalable genome exploration open source tool for variants generated from high-throughput sequencing projects. BMC Bioinformatics, 2019, 20, 61.	1.2	2
1785	Familial chronic megacolon presenting in childhood or adulthood: Seeking the presumed gene association. Neurogastroenterology and Motility, 2019, 31, e13550.	1.6	8
1786	ALG11â€CDG syndrome: Expanding the phenotype. American Journal of Medical Genetics, Part A, 2019, 179, 498-502.	0.7	8
1787	Both rare and common genetic variants contribute to autism in the Faroe Islands. Npj Genomic Medicine, 2019, 4, 1.	1.7	72
1788	Neuropathological correlates and genetic architecture of microglial activation in elderly human brain. Nature Communications, 2019, 10, 409.	5.8	121
1789	Integrative Functional Annotation of 52 Genetic Loci Influencing Myocardial Mass Identifies Candidate Regulatory Variants and Target Genes. Circulation Genomic and Precision Medicine, 2019, 12, e002328.	1.6	7
1790	A case of earlyâ€onset epileptic encephalopathy with a homozygous TBC1D24 variant caused by uniparental isodisomy. American Journal of Medical Genetics, Part A, 2019, 179, 645-649.	0.7	3
1791	Improved Pathogenic Variant Localization via a Hierarchical Model of Sub-regional Intolerance. American Journal of Human Genetics, 2019, 104, 299-309.	2.6	29
1792	Comprehensive functional annotation of susceptibility SNPs prioritized 10 genes for schizophrenia. Translational Psychiatry, 2019, 9, 56.	2.4	20
1793	An essential role for the Zn2+ transporter ZIP7 in B cell development. Nature Immunology, 2019, 20, 350-361.	7.0	92
1794	Genome-wide association studies of impulsive personality traits (BIS-11 and UPPSP) and drug experimentation in up to 22,861 adult research participants identify loci in the CACNA1I and CADM2 genes. Journal of Neuroscience, 2019, 39, 2662-18.	1.7	128
1795	Biallelic pathogenic variants in the lanosterol synthase gene LSS involved in the cholesterol biosynthesis cause alopecia with intellectual disability, a rare recessive neuroectodermal syndrome. Genetics in Medicine, 2019, 21, 2025-2035.	1.1	40
1796	Nonsyndromic craniosynostosis: novel coding variants. Pediatric Research, 2019, 85, 463-468.	1.1	14
1797	Genetic variation in PLEKHG1 is associated with white matter hyperintensities (n = 11,226). Neurology, 2019, 92, e749-e757.	1.5	47
1798	Biological relevance of computationally predicted pathogenicity of noncoding variants. Nature Communications, 2019, 10, 330.	5.8	44
1799	Loss of function BMP4 mutation supports the implication of the BMP/TGFâ€Î² pathway in the etiology of combined pituitary hormone deficiency. American Journal of Medical Genetics, Part A, 2019, 179, 1591-1597.	0.7	9

#	Article	IF	Citations
1800	A familial case of pseudohypoaldosteronism type II (PHA2) with a novel mutation (D564N) in the acidic motif in WNK4. Molecular Genetics & Genomic Medicine, 2019, 7, e705.	0.6	5
1801	Assessment of PARP4 as a candidate breast cancer susceptibility gene. Breast Cancer Research and Treatment, 2019, 177, 145-153.	1.1	8
1802	Predicting disease-causing variant combinations. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 11878-11887.	3.3	68
1803	Innovative strategies for annotating the â€œrelationSNPâ€•between variants and molecular phenotypes. BioData Mining, 2019, 12, 10.	2.2	6
1804	GATA4 screening in Iranian patients of various ethnicities affected with congenital heart disease: Coâ€•occurrence of a novel de novo translocation (5;7) and a likely pathogenic heterozygous GATA4 mutation in a family with autosomal dominant congenital heart disease. Journal of Clinical Laboratory Analysis, 2019, 33, e22923.	0.9	7
1805	Fido-SNP: the first webserver for scoring the impact of single nucleotide variants in the dog genome. Nucleic Acids Research, 2019, 47, W136-W141.	6.5	3
1806	Star Allele-Based Haplotyping versus Gene-Wise Variant Burden Scoring for Predicting 6-Mercaptopurine Intolerance in Pediatric Acute Lymphoblastic Leukemia Patients. Frontiers in Pharmacology, 2019, 10, 654.	1.6	16
1807	Expanded Phenotypic Spectrum of Retinopathies Associated with Autosomal Recessive and Dominant Mutations in PROM1. American Journal of Ophthalmology, 2019, 207, 204-214.	1.7	17
1808	Comprehensive Multiple eQTL Detection and Its Application to GWAS Interpretation. Genetics, 2019, 212, 905-918.	1.2	23
1809	CHASMplus Reveals the Scope of Somatic Missense Mutations Driving Human Cancers. Cell Systems, 2019, 9, 9-23.e8.	2.9	83
1810	Laying the foundation for genomically-based risk assessment in chronic myeloid leukemia. Leukemia, 2019, 33, 1835-1850.	3.3	97
1811	Deleterious de novo variants of Xâ€•linked ZC4H2 in females cause a variable phenotype with neurogenic arthrogryposis multiplex congenita. Human Mutation, 2019, 40, 2270-2285.	1.1	29
1812	Inherited IL-18BP deficiency in human fulminant viral hepatitis. Journal of Experimental Medicine, 2019, 216, 1777-1790.	4.2	70
1813	Exome Sequencing of a Primary Ovarian Insufficiency Cohort Reveals Common Molecular Etiologies for a Spectrum of Disease. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 3049-3067.	1.8	53
1814	Mutation spectrum of Charcotâ€•Marieâ€•Tooth disease among the Han Chinese in Taiwan. Annals of Clinical and Translational Neurology, 2019, 6, 1090-1101.	1.7	29
1815	Characterization of intellectual disability and autism comorbidity through gene panel sequencing. Human Mutation, 2019, 40, 1346-1363.	1.1	54
1816	Frequency and phenotypic spectrum of KMT2B dystonia in childhood: A singleâ€•center cohort study. Movement Disorders, 2019, 34, 1516-1527.	2.2	55
1817	Severe influenza pneumonitis in children with inherited TLR3 deficiency. Journal of Experimental Medicine, 2019, 216, 2038-2056.	4.2	134

#	Article	IF	Citations
1818	An evaluation of noncoding genome annotation tools through enrichment analysis of 15 genome-wide association studies. Briefings in Bioinformatics, 2019, 20, 995-1003.	3.2	3
1819	Decreased ACKR3 (CXCR7) function causes oculomotor synkinesis in mice and humans. Human Molecular Genetics, 2019, 28, 3113-3125.	1.4	8
1820	A rare functional variant of SHARPIN attenuates the inflammatory response and associates with increased risk of late-onset Alzheimerâ€™s disease. Molecular Medicine, 2019, 25, 20.	1.9	33
1821	Incomplete penetrance of CRX gene for autosomal dominant form of cone-rod dystrophy. Ophthalmic Genetics, 2019, 40, 259-266.	0.5	11
1822	Geneâ€specific features enhance interpretation of mutational impact on acid Î±â€glucosidase enzyme activity. Human Mutation, 2019, 40, 1507-1518.	1.1	8
1823	Exome Sequencing in Clinical Hepatology. Hepatology, 2019, 70, 2185-2192.	3.6	19
1824	Predicting functional variants in enhancer and promoter elements using RegulomeDB. Human Mutation, 2019, 40, 1292-1298.	1.1	71
1825	Exome-wide assessment of the functional impact and pathogenicity of multinucleotide mutations. Genome Research, 2019, 29, 1047-1056.	2.4	34
1826	Patterns of genetic structure and adaptive positive selection in the Lithuanian population from high-density SNP data. Scientific Reports, 2019, 9, 9163.	1.6	13
1827	Neutrophil GM-CSF signaling in inflammatory bowel disease patients is influenced by non-coding genetic variants. Scientific Reports, 2019, 9, 9168.	1.6	3
1828	Multiple Sclerosis patients carry an increased burden of exceedingly rare genetic variants in the inflammasome regulatory genes. Scientific Reports, 2019, 9, 9171.	1.6	33
1829	PRRT2 mutations in Japanese patients with benign infantile epilepsy and paroxysmal kinesigenic dyskinesia. Seizure: the Journal of the British Epilepsy Association, 2019, 71, 1-5.	0.9	16
1830	Severe Noonan syndrome phenotype associated with a germline Q71R MRAS variant: a recurrent substitution in RAS homologs in various cancers. American Journal of Medical Genetics, Part A, 2019, 179, 1628-1630.	0.7	13
1831	RAPIDOMICS: rapid genome-wide sequencing in a neonatal intensive care unitâ€”successes and challenges. European Journal of Pediatrics, 2019, 178, 1207-1218.	1.3	59
1832	Germline variants and somatic mutation signatures of breast cancer across populations of African and European ancestry in the US and Nigeria. International Journal of Cancer, 2019, 145, 3321-3333.	2.3	16
1833	The maternal immune activation model uncovers a role for the Arx gene in GABAergic dysfunction in schizophrenia. Brain, Behavior, and Immunity, 2019, 81, 161-171.	2.0	26
1834	Loci for human leukocyte telomere length in the Singaporean Chinese population and trans-ethnic genetic studies. Nature Communications, 2019, 10, 2491.	5.8	64
1835	Identification and in-silico analysis of a novel disease-causing variant in the GUSB gene for Mucopolysaccharidosis VII presenting as non-immune fetal hydrops. Gene Reports, 2019, 16, 100437.	0.4	1

#	Article	IF	Citations
1836	Exome sequencing in multiple sclerosis families identifies 12 candidate genes and nominates biological pathways for the genesis of disease. PLoS Genetics, 2019, 15, e1008180.	1.5	46
1837	Autism and developmental disability caused by KCNQ3 gainâ€ofâ€function variants. Annals of Neurology, 2019, 86, 181-192.	2.8	73
1838	Unravelling the covalent binding of zampanolide and taccalonolide AJ to a minimalist representation of a human microtubule. Journal of Computer-Aided Molecular Design, 2019, 33, 627-644.	1.3	11
1839	A de novo gain-of-function KCND3 mutation in early repolarization syndrome. Heart Rhythm, 2019, 16, 1698-1706.	0.3	30
1840	EFL1 mutations impair eIF6 release to cause Shwachman-Diamond syndrome. Blood, 2019, 134, 277-290.	0.6	48
1841	Association of Early-Onset Alzheimer Disease With Elevated Low-Density Lipoprotein Cholesterol Levels and Rare Genetic Coding Variants of APOB. JAMA Neurology, 2019, 76, 809.	4.5	94
1842	Muscular dystrophy with arrhythmia caused by loss-of-function mutations in BVES. Neurology: Genetics, 2019, 5, e321.	0.9	26
1843	Comprehensive Analysis of Rare Variants of 101 Autism-Linked Genes in a Hungarian Cohort of Autism Spectrum Disorder Patients. Frontiers in Genetics, 2019, 10, 434.	1.1	14
1844	Assessing concordance among human, in silico predictions and functional assays on genetic variant classification. Bioinformatics, 2019, 35, 5163-5170.	1.8	4
1845	Late Jomon male and female genome sequences from the Funadomari site in Hokkaido, Japan. Anthropological Science, 2019, 127, 83-108.	0.2	58
1846	A catalog of genetic loci associated with kidney function from analyses of a million individuals. Nature Genetics, 2019, 51, 957-972.	9.4	549
1847	Noninvasive diagnosis of TRIT1â€related mitochondrial disorder by measuring i⁶A37 and ms²i⁶A37 modifications in tRNAs from blood and urine samples. American Journal of Medical Genetics, Part A, 2019, 179, 1609-1614.	0.7	6
1848	Flexible and scalable diagnostic filtering of genomic variants using G2P with Ensembl VEP. Nature Communications, 2019, 10, 2373.	5.8	86
1849	Rare variant phasing using paired tumor:normal sequence data. BMC Bioinformatics, 2019, 20, 265.	1.2	2
1850	Functional Genetic Variation in the Anti-MÃ¼llerian Hormone Pathway in Women With Polycystic Ovary Syndrome. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 2855-2874.	1.8	58
1851	Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts. Nature Medicine, 2019, 25, 911-919.	15.2	221
1852	Targeted next generation sequencing as a tool for precision medicine. BMC Medical Genomics, 2019, 12, 81.	0.7	54
1853	Structural prediction, whole exome sequencing and molecular dynamics simulation confirms p.G118D somatic mutation of PIK3CA as functionally important in breast cancer patients. Computational Biology and Chemistry, 2019, 80, 472-479.	1.1	5

#	Article	IF	Citations
1854	PEDIA: prioritization of exome data by image analysis. Genetics in Medicine, 2019, 21, 2807-2814.	1.1	58
1855	A novel TBK1 mutation in a family with diverse frontotemporal dementia spectrum disorders. Journal of Physical Education and Sports Management, 2019, 5, a003913.	0.5	19
1856	Phenotype characterisation of TBX4 mutation and deletion carriers with neonatal and paediatric pulmonary hypertension. European Respiratory Journal, 2019, 54, 1801965.	3.1	77
1857	Whole Exome Sequencing Identified a Novel Biallelic SMARCAL1 Mutation in the Extremely Rare Disease SIOD. Frontiers in Genetics, 2019, 10, 565.	1.1	4
1858	Genetic Predisposition for Immune System, Hormone, and Metabolic Dysfunction in Myalgic Encephalomyelitis/Chronic Fatigue Syndrome: A Pilot Study. Frontiers in Pediatrics, 2019, 7, 206.	0.9	24
1859	Largeâ€scale functionalLIPAvariant characterization to improve birth prevalence estimates of lysosomal acid lipase deficiency. Human Mutation, 2019, 40, 2007-2020.	1.1	13
1860	A catalog of single nucleotide changes distinguishing modern humans from archaic hominins. Scientific Reports, 2019, 9, 8463.	1.6	60
1861	Mutational landscape of head and neck squamous cell carcinomas in a South Asian population. Genetics and Molecular Biology, 2019, 42, 526-542.	0.6	6
1862	Peripheral neuropathy and cognitive impairment associated with a novel monoallelic <scp>HARS</scp> variant. Annals of Clinical and Translational Neurology, 2019, 6, 1072-1080.	1.7	15
1863	In trans variant calling reveals enrichment for compound heterozygous variants in genes involved in neuronal development and growth.. Genetical Research, 2019, 101, e8.	0.3	3
1864	Genetic Influences on Behavioral Outcomes After Childhood TBI: A Novel Systems Biology-Informed Approach. Frontiers in Genetics, 2019, 10, 481.	1.1	16
1865	Identification and characterization of rare toll-like receptor 3 variants in patients with autoimmune Addison's disease. Journal of Translational Autoimmunity, 2019, 1, 100005.	2.0	5
1866	Pathogenicity and functional impact of non-frameshifting insertion/deletion variation in the human genome. PLoS Computational Biology, 2019, 15, e1007112.	1.5	34
1867	Primary cilia defects causing mitral valve prolapse. Science Translational Medicine, 2019, 11, .	5.8	76
1868	A personalized platform identifies trametinib plus zoledronate for a patient with KRAS-mutant metastatic colorectal cancer. Science Advances, 2019, 5, eaav6528.	4.7	74
1869	Gene pathogenicity prediction of Mendelian diseases via the random forest algorithm. Human Genetics, 2019, 138, 673-679.	1.8	4
1870	Metabolic reprogramming toward oxidative phosphorylation identifies a therapeutic target for mantle cell lymphoma. Science Translational Medicine, 2019, 11, .	5.8	161
1871	A fullyâ€automated eventâ€based variant prioritizing solution to the CAGI5 intellectual disability gene panel challenge. Human Mutation, 2019, 40, 1364-1372.	1.1	3

#	Article	IF	Citations
1872	Predicting the change of exon splicing caused by genetic variant using support vector regression. Human Mutation, 2019, 40, 1235-1242.	1.1	6
1873	Targeted next generation sequencing in 112 Chinese patients with intellectual disability/developmental delay: novel mutations and candidate gene. BMC Medical Genetics, 2019, 20, 80.	2.1	18
1874	Estimating prevalence for limb-girdle muscular dystrophy based on public sequencing databases. Genetics in Medicine, 2019, 21, 2512-2520.	1.1	56
1875	Integration of multiple epigenomic marks improves prediction of variant impact in saturation mutagenesis reporter assay. Human Mutation, 2019, 40, 1280-1291.	1.1	46
1876	Novel EYA4 variant in Slovak family with late onset autosomal dominant hearing loss: a case report. BMC Medical Genetics, 2019, 20, 84.	2.1	9
1877	BRCA1 â€•and BRCA2 â€šspecific in silico tools for variant interpretation in the CAGI 5 ENIGMA challenge. Human Mutation, 2019, 40, 1593-1611.	1.1	11
1878	Prospective Longitudinal ctDNA Workflow Reveals Clinically Actionable Alterations in Ovarian Cancer. JCO Precision Oncology, 2019, 3, 1-12.	1.5	20
1879	Challenges and Considerations in Sequence Variant Interpretation for Mendelian Disorders. Annals of Laboratory Medicine, 2019, 39, 421-429.	1.2	31
1880	Association of Variants in PINX1 and TREM2 With Late-Onset Alzheimer Disease. JAMA Neurology, 2019, 76, 942.	4.5	20
1881	Removal of alleles by genome editing (RAGE) against deleterious load. Genetics Selection Evolution, 2019, 51, 14.	1.2	44
1882	Using secondary structure to predict the effects of genetic variants on alternative splicing. Human Mutation, 2019, 40, 1270-1279.	1.1	4
1883	Excessive Seizure Clusters in an Otherwise Well-Controlled Epilepsy as a Possible Hallmark of Untreated Vitamin B6-Responsive Epilepsy due to a Homozygous PLPBP Missense Variant. Journal of Pediatric Genetics, 2019, 08, 222-225.	0.3	9
1884	A segregating human allele ofSPO11modeled in mice disrupts timing and amounts of meiotic recombination, causing oligospermia and a decreased ovarian reserveâ€. Biology of Reproduction, 2019, 101, 347-359.	1.2	10
1885	Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis. Human Mutation, 2019, 40, 1115-1126.	1.1	19
1886	Mutational spectrum of dystrophinopathies in Singapore: Insights for genetic diagnosis and precision therapy. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 230-244.	0.7	12
1887	Somatic variants in epigenetic modifiers can predict failure of response to imatinib but not to second-generation tyrosine kinase inhibitors. Haematologica, 2019, 104, 2400-2409.	1.7	37
1888	GenePy - a score for estimating gene pathogenicity in individuals using next-generation sequencing data. BMC Bioinformatics, 2019, 20, 254.	1.2	21
1889	Predisposing germline mutations in high hyperdiploid acute lymphoblastic leukemia in children. Genes Chromosomes and Cancer, 2019, 58, 723-730.	1.5	17

#	Article	IF	Citations
1890	Pharmacogenomics of Cisplatinâ€Induced Ototoxicity: Successes, Shortcomings, and Future Avenues of Research. Clinical Pharmacology and Therapeutics, 2019, 106, 350-359.	2.3	24
1891	Study of chromatin remodeling genes implicates SMARCA4 as a putative player in oncogenesis in neuroblastoma. International Journal of Cancer, 2019, 145, 2781-2791.	2.3	16
1892	Rare variants in MYH15 modify amyotrophic lateral sclerosis risk. Human Molecular Genetics, 2019, 28, 2309-2318.	1.4	4
1893	Homozygous noncanonical splice variant in LSM1 in two siblings with multiple congenital anomalies and global developmental delay. Journal of Physical Education and Sports Management, 2019, 5, a004101.	0.5	4
1894	Identification of novel mutations in preaxial polydactyly patients through wholeâ€exome sequencing. Molecular Genetics & Genomic Medicine, 2019, 7, e690.	0.6	7
1895	Functional characterization of 3D protein structures informed by human genetic diversity. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 8960-8965.	3.3	33
1896	Early-onset emphysema in a large French-Canadian family: a genetic investigation. Lancet Respiratory Medicine,the, 2019, 7, 427-436.	5.2	15
1897	A CCR4-NOT Transcription Complex, Subunit 1, CNOT1, Variant Associated with Holoprosencephaly. American Journal of Human Genetics, 2019, 104, 990-993.	2.6	30
1898	Human genotyping and an experimental model reveal NPR-C as a possible contributor to morbidity in coarctation of the aorta. Physiological Genomics, 2019, 51, 177-185.	1.0	12
1899	Towards controlled terminology for reporting germline cancer susceptibility variants: an ENIGMA report. Journal of Medical Genetics, 2019, 56, 347-357.	1.5	32
1900	Genetic Techniques Used in the Diagnosis of Inherited Platelet Disorders. Seminars in Thrombosis and Hemostasis, 2019, 45, 685-694.	1.5	4
1901	TAGOOS: genome-wide supervised learning of non-coding loci associated to complex phenotypes. Nucleic Acids Research, 2019, 47, e79-e79.	6.5	3
1902	Whole genome sequencing and variant discovery in the ASPIRE autism spectrum disorder cohort. Clinical Genetics, 2019, 96, 199-206.	1.0	18
1903	Interchromosomal template-switching as a novel molecular mechanism for imprinting perturbations associated with Temple syndrome. Genome Medicine, 2019, 11, 25.	3.6	22
1904	SeqTailor: a user-friendly webserver for the extraction of DNA or protein sequences from next-generation sequencing data. Nucleic Acids Research, 2019, 47, W623-W631.	6.5	15
1905	Mitochondrial Disease Genetics. , 2019, , 41-62.		0
1906	Variants in myelin regulatory factor (MYRF) cause autosomal dominant and syndromic nanophthalmos in humans and retinal degeneration in mice. PLoS Genetics, 2019, 15, e1008130.	1.5	50
1907	DNAscan: personal computer compatible NGS analysis, annotation and visualisation. BMC Bioinformatics, 2019, 20, 213.	1.2	14

#	Article	IF	Citations
1908	Targeted NGS Platforms for Genetic Screening and Gene Discovery in Primary Immunodeficiencies. Frontiers in Immunology, 2019, 10, 316.	2.2	42
1909	Minimal inflammatory foci of unknown etiology may be a tentative sign of early stage inherited cardiomyopathy. Modern Pathology, 2019, 32, 1281-1290.	2.9	16
1910	FunSPU: A versatile and adaptive multiple functional annotation-based association test of whole-genome sequencing data. PLoS Genetics, 2019, 15, e1008081.	1.5	16
1911	Anti-Hypothalamus and Anti-Pituitary AutoÂantibodies in ROHHAD Syndrome: Additional Evidence Supporting an Autoimmune Etiopathogenesis. Hormone Research in Paediatrics, 2019, 92, 124-132.	0.8	27
1912	Translating genomics to the clinical diagnosis of disorders/differences of sex development. Current Topics in Developmental Biology, 2019, 134, 317-375.	1.0	25
1913	Shared genetic underpinnings of childhood obesity and adult cardiometabolic diseases. Human Genomics, 2019, 13, 17.	1.4	17
1914	Mouse screen reveals multiple new genes underlying mouse and human hearing loss. PLoS Biology, 2019, 17, e3000194.	2.6	84
1915	Improved measures for evolutionary conservation that exploit taxonomy distances. Nature Communications, 2019, 10, 1556.	5.8	21
1916	Novel likely pathogenic variants in TMEM126A identified in non-syndromic autosomal recessive optic atrophy: two case reports. BMC Medical Genetics, 2019, 20, 62.	2.1	11
1917	Linkage analysis and whole exome sequencing reveals AHNAK2 as a novel genetic cause for autosomal recessive CMT in a Malaysian family. Neurogenetics, 2019, 20, 117-127.	0.7	12
1918	Variant Score Rankerâ€"a web application for intuitive missense variant prioritization. Bioinformatics, 2019, 35, 4478-4479.	1.8	5
1919	eDiVAâ€"Classification and prioritization of pathogenic variants for clinical diagnostics. Human Mutation, 2019, 40, 865-878.	1.1	19
1920	Rare and Low-Frequency Variants in RNF213 Confer Susceptibility to Moyamoya Syndrome Associated with Hyperthyroidism. World Neurosurgery, 2019, 127, e460-e466.	0.7	5
1921	Novel mutations in MYBPC1 are associated with myogenic tremor and mild myopathy. Annals of Neurology, 2019, 86, 129-142.	2.8	27
1922	Gaps in Current Autism Research: The Thoughts of the Autism Research Editorial Board and Associate Editors. Autism Research, 2019, 12, 700-714.	2.1	28
1923	A high prevalence of biallelicRPE65mutations in Costa Rican children with Leber congenital amaurosis and early-onset retinal dystrophy. Ophthalmic Genetics, 2019, 40, 110-117.	0.5	7
1924	Panel-Based Nuclear and Mitochondrial Next-Generation Sequencing Outcomes of an Ethnically Diverse Pediatric Patient Cohort with Mitochondrial Disease. Journal of Molecular Diagnostics, 2019, 21, 503-513.	1.2	12
1925	Pathogenetic basis of Takenouchi-Kosaki syndrome: Electron microscopy study using platelets in patients and functional studies in a Caenorhabditis elegans model. Scientific Reports, 2019, 9, 4418.	1.6	16

#	Article	IF	Citations
1926	Integrated Analysis of Whole Exome Sequencing and Copy Number Evaluation in Parkinsonâ€™s Disease. Scientific Reports, 2019, 9, 3344.	1.6	31
1927	ALSgeneScanner: a pipeline for the analysis and interpretation of DNA sequencing data of ALS patients. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2019, 20, 207-215.	1.1	11
1928	UniProt genomic mapping for deciphering functional effects of missense variants. Human Mutation, 2019, 40, 694-705.	1.1	29
1929	A novel nonsense variant in SUPT20H gene associated with Rheumatoid Arthritis identified by Whole Exome Sequencing of multiplex families. PLoS ONE, 2019, 14, e0213387.	1.1	7
1930	MMSplice: modular modeling improves the predictions of genetic variant effects on splicing. Genome Biology, 2019, 20, 48.	3.8	140
1931	SMAD4 rare variants in individuals and families with thoracic aortic aneurysms and dissections. European Journal of Human Genetics, 2019, 27, 1054-1060.	1.4	24
1932	A pathway-driven predictive model of tramadol pharmacogenetics. European Journal of Human Genetics, 2019, 27, 1143-1156.	1.4	4
1933	Mutations in DNA repair genes are associated with increased neoantigen burden and a distinct immunophenotype in lung squamous cell carcinoma. Scientific Reports, 2019, 9, 3235.	1.6	60
1934	Selecting variants of unknown significance through network-based gene-association significantly improves risk prediction for disease-control cohorts. Scientific Reports, 2019, 9, 3266.	1.6	18
1935	NBAS pathogenic variants: Defining the associated clinical and facial phenotype and genotypeâ€“phenotype correlations. Human Mutation, 2019, 40, 721-728.	1.1	26
1936	Niemann-Pick Disease Type C: Mutation Spectrum and Novel Sequence Variations in the Human NPC1 Gene. Molecular Neurobiology, 2019, 56, 6426-6435.	1.9	15
1937	A familial congenital heart disease with a possible multigenic origin involving a mutation in BMPR1A. Scientific Reports, 2019, 9, 2959.	1.6	14
1938	Phenotype and response to growth hormone therapy in siblings with B4GALT7 deficiency. Bone, 2019, 124, 14-21.	1.4	9
1939	Genomic Analysis in the Age of Human Genome Sequencing. Cell, 2019, 177, 70-84.	13.5	205
1940	Novel compound mutations in the mitochondrial translation elongation factor (TSFM) gene cause severe cardiomyopathy with myocardial fibro-adipose replacement. Scientific Reports, 2019, 9, 5108.	1.6	12
1941	De novo and biallelic DEAF1 variants cause a phenotypic spectrum. Genetics in Medicine, 2019, 21, 2059-2069.	1.1	20
1942	Rare Pathogenic Variants Predispose to Hepatocellular Carcinoma in Nonalcoholic Fatty Liver Disease. Scientific Reports, 2019, 9, 3682.	1.6	85
1943	Deletions and loss-of-function variants in TP63 associated with orofacial clefting. European Journal of Human Genetics, 2019, 27, 1101-1112.	1.4	16

#	Article	IF	Citations
1944	Genome-wide association study in frontal fibrosing alopecia identifies four susceptibility loci including HLA-B*07:02. Nature Communications, 2019, 10, 1150.	5.8	82
1945	De novo missense variant in the GTPase effector domain (GED) of DNM1L leads to static encephalopathy and seizures. Journal of Physical Education and Sports Management, 2019, 5, a003673.	0.5	24
1946	Role of protein structure in variant annotation: structural insight of mutations causing 6-pyruvoyl-tetrahydropterin synthase deficiency. Pathology, 2019, 51, 274-280.	0.3	7
1947	Carrier frequency estimation of Zellweger spectrum disorder using ExAC database and bioinformatics tools. Genetics in Medicine, 2019, 21, 1969-1976.	1.1	10
1948	Molecular basis of familial adenomatous polyposis in the southeast of Brazil: identification of six novel mutations. International Journal of Biological Markers, 2019, 34, 80-89.	0.7	6
1949	Novel mutation in the MED23 gene for intellectual disability: A case report and literature review. Clinical Case Reports (discontinued), 2019, 7, 331-335.	0.2	17
1950	A novel germline mutation of the SFTPA1 gene in familial interstitial pneumonia. Human Genome Variation, 2019, 6, 12.	0.4	15
1951	New insights into the pathogenicity of non-synonymous variants through multi-level analysis. Scientific Reports, 2019, 9, 1667.	1.6	40
1952	Genetic Variation in Pan Species Is Shaped by Demographic History and Harbors Lineage-Specific Functions. Genome Biology and Evolution, 2019, 11, 1178-1191.	1.1	15
1953	mGAP: the macaque genotype and phenotype resource, a framework for accessing and interpreting macaque variant data, and identifying new models of human disease. BMC Genomics, 2019, 20, 176.	1.2	26
1954	Comparison of different functional prediction scores using a gene-based permutation model for identifying cancer driver genes. BMC Medical Genomics, 2019, 12, 22.	0.7	12
1955	How good are pathogenicity predictors in detecting benign variants?. PLoS Computational Biology, 2019, 15, e1006481.	1.5	79
1956	Complications of whole-exome sequencing for causal gene discovery in primary platelet secretion defects. Haematologica, 2019, 104, 2084-2090.	1.7	9
1957	The Frog Xenopus as a Model to Study Joubert Syndrome: The Case of a Human Patient With Compound Heterozygous Variants in PIBF1. Frontiers in Physiology, 2019, 10, 134.	1.3	13
1958	Loss of DPP6 in neurodegenerative dementia: a genetic player in the dysfunction of neuronal excitability. Acta Neuropathologica, 2019, 137, 901-918.	3.9	37
1959	Methods for the Analysis and Interpretation for Rare Variants Associated with Complex Traits. Current Protocols in Human Genetics, 2019, 101, e83.	3.5	11
1960	A Syndromic Neurodevelopmental Disorder Caused by Mutations in SMARCD1, a Core SWI/SNF Subunit Needed for Context-Dependent Neuronal Gene Regulation in Flies. American Journal of Human Genetics, 2019, 104, 596-610.	2.6	32
1961	Deleterious Variation in BRSK2 Associates with a Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 701-708.	2.6	19

#	Article	IF	Citations
1962	Investigation of novel variations of ORAI1 gene and their association with Kawasaki disease. Journal of Human Genetics, 2019, 64, 511-519.	1.1	9
1963	Genetic overlap between birthweight and adult cardiometabolic diseases has implications for genomic medicine. Scientific Reports, 2019, 9, 4076.	1.6	5
1964	ACAT: A Fast and Powerful p Value Combination Method for Rare-Variant Analysis in Sequencing Studies. American Journal of Human Genetics, 2019, 104, 410-421.	2.6	219
1965	The lysosomal storage disorders mucolipidosis type II, type III alpha/beta, and type III gamma: Update onGNPTABandGNPTGmutations. Human Mutation, 2019, 40, 842-864.	1.1	36
1966	Exome Sequencing of Two Siblings with Sporadic Autism Spectrum Disorder and Severe Speech Sound Disorder Suggests Pleiotropic and Complex Effects. Behavior Genetics, 2019, 49, 399-414.	1.4	18
1967	Mutation and association analyses of dementia-causal genes in Han Chinese patients with early-onset and familial Alzheimer's disease. Journal of Psychiatric Research, 2019, 113, 141-147.	1.5	20
1968	Tau aggregation and seeding analyses of two novel MAPT variants found in patients with motor neuron disease and progressive parkinsonism. Neurobiology of Aging, 2019, 84, 240.e13-240.e22.	1.5	10
1969	Big data in der Diagnostik genetischer SchwerhÃ¶rigkeit. Laryngo- Rhino- Otologie, 2019, 98, S32-S81.	0.2	6
1970	Loss of Cajal bodies in motor neurons from patients with novel mutations in VRK1. Human Molecular Genetics, 2019, 28, 2378-2394.	1.4	17
1971	Germline-Derived Gain-of-Function Variants of GsÎ±-Coding GNAS Gene Identified in Nephrogenic Syndrome of Inappropriate Antidiuresis. Journal of the American Society of Nephrology: JASN, 2019, 30, 877-889.	3.0	21
1972	A Combined in silico, in vitro and Clinical Approach to Characterize Novel Pathogenic Missense Variants in PRPF31 in Retinitis Pigmentosa. Frontiers in Genetics, 2019, 10, 248.	1.1	7
1973	Characterizing variants of unknown significance in rhodopsin: A functional genomics approach. Human Mutation, 2019, 40, 1127-1144.	1.1	22
1974	Pathogenic Germ Line Variants in a Patient With Severe Toxicity From Breast Radiotherapy. Clinical Breast Cancer, 2019, 19, e400-e405.	1.1	1
1975	Population-based analysis of BAP1 germline variations in patients with uveal melanoma. Human Molecular Genetics, 2019, 28, 2415-2426.	1.4	17
1976	Genome-wide association study of suicide attempt in a Mexican population: a study protocol. BMJ Open, 2019, 9, e025335.	0.8	2
1977	A patient with homozygous nonsense variants in two Leigh syndrome disease genes: Distinguishing a dual diagnosis from a hypomorphic proteinâ€truncating variant. Human Mutation, 2019, 40, 893-898.	1.1	8
1978	Variants in KIAA0825 underlie autosomal recessive postaxial polydactyly. Human Genetics, 2019, 138, 593-600.	1.8	16
1979	Biallelic variants in SMAD6 are associated with a complex cardiovascular phenotype. Human Genetics, 2019, 138, 625-634.	1.8	12

#	Article	IF	Citations
1980	Rare variants in SLC5A10 are associated with serum 1,5-anhydroglucitol (1,5-AG) in the Atherosclerosis Risk in Communities (ARIC) Study. Scientific Reports, 2019, 9, 5941.	1.6	9
1981	Clinicopathological and Genetic Profiles of Cases with Myocytes Disarrayâ€"Investigation for Establishing the Autopsy Diagnostic Criteria for Hypertrophic Cardiomyopathy. Journal of Clinical Medicine, 2019, 8, 463.	1.0	11
1983	Dynamic Scan Procedure for Detecting Rare-Variant Association Regions in Whole-Genome Sequencing Studies. American Journal of Human Genetics, 2019, 104, 802-814.	2.6	43
1984	Review: Precision medicine and driver mutations: Computational methods, functional assays and conformational principles for interpreting cancer drivers. PLoS Computational Biology, 2019, 15, e1006658.	1.5	83
1985	Mutation p.R356Q in the Collybistin Phosphoinositide Binding Site Is Associated With Mild Intellectual Disability. Frontiers in Molecular Neuroscience, 2019, 12, 60.	1.4	10
1986	De Novo and Inherited Pathogenic Variants in KDM3B Cause Intellectual Disability, Short Stature, and Facial Dysmorphism. American Journal of Human Genetics, 2019, 104, 758-766.	2.6	34
1987	Structural and Computational Characterization of Disease-Related Mutations Involved in Protein-Protein Interfaces. International Journal of Molecular Sciences, 2019, 20, 1583.	1.8	17
1988	Whole-exome sequencing detects mutations in pediatric patients with atypical hemolytic uremic syndrome in Taiwan. Clinica Chimica Acta, 2019, 494, 143-150.	0.5	8
1989	The prevalence of GALM mutations that cause galactosemia: A database of functionally evaluated variants. Molecular Genetics and Metabolism, 2019, 126, 362-367.	0.5	19
1990	Approaches to functionally validate candidate genetic variants involved in colorectal cancer predisposition. Molecular Aspects of Medicine, 2019, 69, 27-40.	2.7	5
1991	Functional and structural analysis of rare SLC2A2 variants associated with Fanconiâ€Bickel syndrome and metabolic traits. Human Mutation, 2019, 40, 983-995.	1.1	13
1992	Genome sequencing for rightward hemispheric language dominance. Genes, Brain and Behavior, 2019, 18, e12572.	1.1	14
1993	Genotype-Phenotype Correlation in Long-Term Cohort of Japanese Patients with Moyamoya Disease. Cerebrovascular Diseases, 2019, 47, 105-111.	0.8	26
1994	MYRF haploinsufficiency causes 46,XY and 46,XX disorders of sex development: bioinformatics consideration. Human Molecular Genetics, 2019, 28, 2319-2329.	1.4	25
1995	Molecular yield of targeted sequencing for Glanzmann thrombasthenia patients. Npj Genomic Medicine, 2019, 4, 4.	1.7	9
1996	DIAPH2 alterations increase cellular motility and may contribute to the metastatic potential of laryngeal squamous cell carcinoma. Carcinogenesis, 2019, 40, 1251-1259.	1.3	12
1997	Confirmation of the role of pathogenic SMAD6 variants in bicuspid aortic valve-related aortopathy. European Journal of Human Genetics, 2019, 27, 1044-1053.	1.4	32
1998	Doubts about TMEM230 as a gene for parkinsonism. Nature Genetics, 2019, 51, 367-368.	9.4	11

#	Article	IF	Citations
1999	Contribution of ultrarare variants in mTOR pathway genes to sporadic focal epilepsies. Annals of Clinical and Translational Neurology, 2019, 6, 475-485.	1.7	15
2000	CVID-Associated Tumors: Czech Nationwide Study Focused on Epidemiology, Immunology, and Genetic Background in a Cohort of Patients With CVID. Frontiers in Immunology, 2018, 9, 3135.	2.2	45
2001	A monoallelic activating mutation in RAC2 resulting in a combined immunodeficiency. Journal of Allergy and Clinical Immunology, 2019, 143, 1649-1653.e3.	1.5	37
2002	Increased frequency of rare missense PPP1R3B variants among Danish patients with type 2 diabetes. PLoS ONE, 2019, 14, e0210114.	1.1	11
2003	Burkitt-like lymphoma with 11q aberration: a germinal center-derived lymphoma genetically unrelated to Burkitt lymphoma. Haematologica, 2019, 104, 1822-1829.	1.7	71
2004	Mutant KCNJ3 and KCNJ5 Potassium Channels as Novel Molecular Targets in Bradyarrhythmias and Atrial Fibrillation. Circulation, 2019, 139, 2157-2169.	1.6	51
2005	The underacknowledged PPA-ALS. Neurology, 2019, 92, e1354-e1366.	1.5	29
2006	The genetic basis of Turner syndrome aortopathy. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 101-109.	0.7	24
2007	Functional genomics reveal gene regulatory mechanisms underlying schizophrenia risk. Nature Communications, 2019, 10, 670.	5.8	94
2008	DeepPVP: phenotype-based prioritization of causative variants using deep learning. BMC Bioinformatics, 2019, 20, 65.	1.2	49
2009	Biallelic Variants in the Nuclear Pore Complex Protein NUP93 Are Associated with Non-progressive Congenital Ataxia. Cerebellum, 2019, 18, 422-432.	1.4	10
2010	Mutational analysis of CFTR in the Ecuadorian population using next-generation sequencing. Gene, 2019, 696, 28-32.	1.0	7
2012	The mutational and phenotypic spectrum of TUBA1A-associated tubulinopathy. Orphanet Journal of Rare Diseases, 2019, 14, 38.	1.2	48
2013	FURINvariant associations with postexercise hypotension are intensity and race dependent. Physiological Reports, 2019, 7, e13952.	0.7	7
2014	Genome-wide analysis of insomnia in 1,331,010 individuals identifies new risk loci and functional pathways. Nature Genetics, 2019, 51, 394-403.	9.4	593
2015	S-CAP extends pathogenicity prediction to genetic variants that affect RNA splicing. Nature Genetics, 2019, 51, 755-763.	9.4	56
2016	Using association signal annotations to boost similarity network fusion. Bioinformatics, 2019, 35, 3718-3726.	1.8	20
2017	Phosphatidylinositol-3-kinase (PI3K)/Akt Signaling is Functionally Essential in Myxoid Liposarcoma. Molecular Cancer Therapeutics, 2019, 18, 834-844.	1.9	28

#	Article	IF	Citations
2018	NCBoost classifies pathogenic non-coding variants in Mendelian diseases through supervised learning on purifying selection signals in humans. Genome Biology, 2019, 20, 32.	3.8	47
2019	Mutations in the Glycosyltransferase Domain of GLT8D1 Are Associated with Familial Amyotrophic Lateral Sclerosis. Cell Reports, 2019, 26, 2298-2306.e5.	2.9	57
2020	Identification of a novel homozygous variant confirms ITPA as a developmental and epileptic encephalopathy gene. American Journal of Medical Genetics, Part A, 2019, 179, 857-861.	0.7	14
2021	SLC35A2â€CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. Human Mutation, 2019, 40, 908-925.	1.1	39
2022	FAM92A Underlies Nonsyndromic Postaxial Polydactyly in Humans and an Abnormal Limb and Digit Skeletal Phenotype in Mice. Journal of Bone and Mineral Research, 2019, 34, 375-386.	3.1	27
2023	Exome sequencing in 51 early onset nonâ€familial CRC cases. Molecular Genetics & Genomic Medicine, 2019, 7, e605.	0.6	17
2024	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	2.6	30
2025	Genetic meta-analysis of diagnosed Alzheimerâ€™s disease identifies new risk loci and implicates AÎ², tau, immunity and lipid processing. Nature Genetics, 2019, 51, 414-430.	9.4	1,962
2026	Comprehensive sequencing of the myocilin gene in a selected cohort of severe primary open-angle glaucoma patients. Scientific Reports, 2019, 9, 3100.	1.6	8
2027	A Novel Rare Missense Variation of the NOD2 Gene: Evidencesof Implication in Crohnâ€™s Disease. International Journal of Molecular Sciences, 2019, 20, 835.	1.8	7
2028	Germline DNA Repair Gene Mutations in Young-onset Prostate Cancer Cases in the UK: Evidence for a More Extensive Genetic Panel. European Urology, 2019, 76, 329-337.	0.9	48
2029	Whole Genome Sequence, Variant Discovery and Annotation in Mapuche-Huilliche Native South Americans. Scientific Reports, 2019, 9, 2132.	1.6	12
2030	Discussion of â€˜Gene hunting with hidden Markov model knockoffsâ€™. Biometrika, 2019, 106, 23-26.	1.3	3
2031	CERENKOV2: improved detection of functional noncoding SNPs using data-space geometric features. BMC Bioinformatics, 2019, 20, 63.	1.2	5
2032	Role of Segregation for Variant Discovery in Multiplex Families Ascertained by Probands With Left Sided Cardiovascular Malformations. Frontiers in Genetics, 2019, 9, 729.	1.1	4
2033	Novel mutations in the RS1 gene in Japanese patients with X-linked congenital retinoschisis. Human Genome Variation, 2019, 6, 3.	0.4	18
2034	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	2.6	27
2035	The Oculome Panel Test. Ophthalmology, 2019, 126, 888-907.	2.5	77

#	Article	IF	Citations
2036	Predicting Pathogenic Non-coding Variants on Imbalanced Data Set using Cluster Ensemble Sampling. , 2019, , .		2
2037	Integrative Modeling and Novel Technologies in Human Genomics. , 2019, , 155-189.		0
2038	Biopsy of palliative lesions following radiotherapy. BJR|Open, 2019, 1, 20180025.	0.4	0
2039	Identification of an Endoglin Variant Associated With HCV-Related Liver Fibrosis Progression by Next-Generation Sequencing. Frontiers in Genetics, 2019, 10, 1024.	1.1	6
2040	Frequent germline mutations of HAVCR2 in sporadic subcutaneous panniculitis-like T-cell lymphoma. Blood Advances, 2019, 3, 588-595.	2.5	73
2041	Advances in identifying coding variants of common complex diseases. Journal of Bio-X Research, 2019, 2, 153-158.	0.3	0
2042	Refinement of evolutionary medicine predictions based on clinical evidence for the manifestations of Mendelian diseases. Scientific Reports, 2019, 9, 18577.	1.6	7
2043	A Recurrent Rare SOX9 Variant (M469V) is Associated with Congenital Vertebral Malformations. Current Gene Therapy, 2019, 19, 242-247.	0.9	11
2044	QTG-Finder: A Machine-Learning Based Algorithm To Prioritize Causal Genes of Quantitative Trait Loci in Arabidopsis and Rice. G3: Genes, Genomes, Genetics, 2019, 9, 3129-3138.	0.8	21
2045	Leveraging protein dynamics to identify cancer mutational hotspots using 3D structures. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 18962-18970.	3.3	26
2046	In Silico Assessment of the Structural, Functional and Stability Impact of a Nonsense PRF1 Mutation with Uncertain Clinical Significance; Identified in 2 Unrelated Cypriot Triple-Negative Breast Cancer Patients. , 2019, , .		1
2047	Genetics of Congenital Isolated TSH Deficiency: Mutation Screening of the Known Causative Genes and a Literature Review. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 6229-6237.	1.8	15
2048	Clinical and genetic variability in children with partial albinism. Scientific Reports, 2019, 9, 16576.	1.6	26
2049	How common are single gene mutations as a cause for lacunar stroke?. Neurology, 2019, 93, e2007-e2020.	1.5	26
2050	Novel risk genes and mechanisms implicated by exome sequencing of 2572 individuals with pulmonary arterial hypertension. Genome Medicine, 2019, 11, 69.	3.6	86
2051	A meta-analysis of genome-wide association studies of epigenetic age acceleration. PLoS Genetics, 2019, 15, e1008104.	1.5	83
2052	Compound heterozygous mutations in SNAP29 is associated with Pelizaeus-Merzbacher-like disorder (PMLD). Human Genetics, 2019, 138, 1409-1417.	1.8	14
2053	Ranking of non-coding pathogenic variants and putative essential regions of the human genome. Nature Communications, 2019, 10, 5241.	5.8	65

#	Article	IF	Citations
2054	Application of Computational Biology and Artificial Intelligence Technologies in Cancer Precision Drug Discovery. BioMed Research International, 2019, 2019, 1-15.	0.9	42
2055	DNA variants affecting the expression of numerous genes in trans have diverse mechanisms of action and evolutionary histories. PLoS Genetics, 2019, 15, e1008375.	1.5	34
2056	Whole-Genome Methylation Analysis of Phenotype Discordant Monozygotic Twins Reveals Novel Epigenetic Perturbation Contributing to the Pathogenesis of Adolescent Idiopathic Scoliosis. Frontiers in Bioengineering and Biotechnology, 2019, 7, 364.	2.0	17
2057	Association of interleukin 22 receptor subunit alpha 1 gene polymorphisms with chronic rhinosinusitis. Brazilian Journal of Otorhinolaryngology, 2021, 87, 505-511.	0.4	3
2058	The Use of Whole Exome Sequencing in a Cohort of Transgender Individuals to Identify Rare Genetic Variants. Scientific Reports, 2019, 9, 20099.	1.6	18
2059	Population size influences the type of nucleotide variations in humans. BMC Genetics, 2019, 20, 93.	2.7	2
2060	Ontology-based prediction of cancer driver genes. Scientific Reports, 2019, 9, 17405.	1.6	16
2061	Chronic mucocutaneous candidiasis and connective tissue disorder in humans with impaired JNK1-dependent responses to IL-17A/F and TGF-Î². Science Immunology, 2019, 4, .	5.6	45
2062	Germline de novo variants in CSNK2B in Chinese patients with epilepsy. Scientific Reports, 2019, 9, 17909.	1.6	26
2063	Artificial intelligence in clinical and genomic diagnostics. Genome Medicine, 2019, 11, 70.	3.6	205
2064	Personalised analytics for rare disease diagnostics. Nature Communications, 2019, 10, 5274.	5.8	15
2065	Homozygous Null TBX4 Mutations Lead to Posterior Amelia with Pelvic and Pulmonary Hypoplasia. American Journal of Human Genetics, 2019, 105, 1294-1301.	2.6	17
2066	Novel SAMD9 Mutation in a Patient With Immunodeficiency, Neutropenia, Impaired Anti-CMV Response, and Severe Gastrointestinal Involvement. Frontiers in Immunology, 2019, 10, 2194.	2.2	12
2067	Biological and practical implications of genome-wide association study of schizophrenia using Bayesian variable selection. NPJ Schizophrenia, 2019, 5, 19.	2.0	0
2068	Sex-Based Analysis of De Novo Variants in Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 105, 1274-1285.	2.6	84
2069	Fine-Mapping Array Design for Multi-Ethnic Studies of Multiple Sclerosis. Genes, 2019, 10, 903.	1.0	3
2070	Clinical Spectrum and Functional Consequences Associated with Bi-Allelic Pathogenic PNPT1 Variants. Journal of Clinical Medicine, 2019, 8, 2020.	1.0	16
2071	VPOT: A Customizable Variant Prioritization Ordering Tool for Annotated Variants. Genomics, Proteomics and Bioinformatics, 2019, 17, 540-545.	3.0	10

#	Article	IF	Citations
2072	Identification of genetic overlap and novel risk loci for attention-deficit/hyperactivity disorder and bipolar disorder. Molecular Psychiatry, 2021, 26, 4055-4065.	4.1	31
2073	Targeted resequencing identifies genes with recurrent variation in cerebral palsy. Npj Genomic Medicine, 2019, 4, 27.	1.7	22
2074	Human SNORA31 variations impair cortical neuron-intrinsic immunity to HSV-1 and underlie herpes simplex encephalitis. Nature Medicine, 2019, 25, 1873-1884.	15.2	76
2075	RegSNPs-intron: a computational framework for predicting pathogenic impact of intronic single nucleotide variants. Genome Biology, 2019, 20, 254.	3.8	52
2076	An AARS variant as the likely cause of Swedish type hereditary diffuse leukoencephalopathy with spheroids. Acta Neuropathologica Communications, 2019, 7, 188.	2.4	19
2077	Machine Learning Approaches for the Prioritization of Genomic Variants Impacting Pre-mRNA Splicing. Cells, 2019, 8, 1513.	1.8	41
2078	First description of the molecular and clinical characterization of hereditary factor V deficiency in Saudi Arabia. Blood Coagulation and Fibrinolysis, 2019, 30, 224-232.	0.5	5
2079	Effects of Common and Rare Chromosome 4 GABAergic Gene Variation on Alcohol Use and Antisocial Behavior. Journal of Studies on Alcohol and Drugs, 2019, 80, 585-593.	0.6	0
2080	From Clinical Phenotype to Genotypic Modelling: Incidence and Prevalence of Recessive Dystrophic Epidermolysis Bullosa (RDEB) . Clinical, Cosmetic and Investigational Dermatology, 2019, Volume 12, 933-942.	0.8	15
2081	Computational and Pharmacogenomic Insights on Hypertension Treatment: Rational Drug Design and Optimization Strategies. Current Drug Targets, 2019, 21, 18-33.	1.0	6
2082	Single Molecule Molecular Inversion Probes for High Throughput Germline Screenings in Dystonia. Frontiers in Neurology, 2019, 10, 1332.	1.1	2
2083	PsyMuKB: An Integrative De Novo Variant Knowledge Base for Developmental Disorders. Genomics, Proteomics and Bioinformatics, 2019, 17, 453-464.	3.0	10
2084	Genetic variations in olfactory receptor gene OR2AG2 in a large multigenerational family with asthma. Scientific Reports, 2019, 9, 19029.	1.6	12
2085	Genome sequencing for early-onset or atypical dementia: high diagnostic yield and frequent observation of multiple contributory alleles. Journal of Physical Education and Sports Management, 2019, 5, a003491.	0.5	25
2086	Whole genome DNA sequencing provides an atlas of somatic mutagenesis in healthy human cells and identifies a tumor-prone cell type. Genome Biology, 2019, 20, 285.	3.8	46
2087	Mutually reinforcing effects of genetic variants and interferonâ€Î² 1a therapy for pulmonary arterial hypertension development in multiple sclerosis patients. Pulmonary Circulation, 2019, 9, 1-6.	0.8	9
2088	Recent genetic and functional insights in autism spectrum disorder. Current Opinion in Neurology, 2019, 32, 627-634.	1.8	7
2089	The kidney transcriptome, from single cells to whole organs and back. Current Opinion in Nephrology and Hypertension, 2019, 28, 219-226.	1.0	11

#	Article	IF	Citations
2090	Genomic Relationships, Novel Loci, and Pleiotropic Mechanisms across Eight Psychiatric Disorders. Cell, 2019, 179, 1469-1482.e11.	13.5	935
2091	The molecular landscape of ETMR at diagnosis and relapse. Nature, 2019, 576, 274-280.	13.7	94
2092	Using mechanistic models for the clinical interpretation of complex genomic variation. Scientific Reports, 2019, 9, 18937.	1.6	20
2093	Integrating hypertension phenotype and genotype with hybrid non-negative matrix factorization. Bioinformatics, 2019, 35, 1395-1403.	1.8	12
2094	IL-17A is functionally relevant and a potential therapeutic target in bullous pemphigoid. Journal of Autoimmunity, 2019, 96, 104-112.	3.0	85
2095	Variations in maternal adenylate cyclase genes are associated with congenital Zika syndrome in a cohort from Northeast, Brazil. Journal of Internal Medicine, 2019, 285, 215-222.	2.7	18
2096	Rare variants in the genetic background modulate cognitive and developmental phenotypes in individuals carrying disease-associated variants. Genetics in Medicine, 2019, 21, 816-825.	1.1	127
2097	PROKR2mutations in idiopathic hypogonadotropic hypogonadism: selective disruption of the binding to aGl±â€protein leads to biased signaling. FASEB Journal, 2019, 33, 4538-4546.	0.2	18
2098	Identification of a Loss-of-Function Mutation in the Context of Glutaminase Deficiency and Neonatal Epileptic Encephalopathy. JAMA Neurology, 2019, 76, 342.	4.5	33
2099	Nonâ€leukemic pediatric mixed phenotype acute leukemia/lymphoma: Genomic characterization and clinical outcome in a prospective trial for pediatric lymphoblastic lymphoma. Genes Chromosomes and Cancer, 2019, 58, 365-372.	1.5	6
2100	Cryptic intronic NBAS variant reveals the genetic basis of recurrent liver failure in a child. Molecular Genetics and Metabolism, 2019, 126, 77-82.	0.5	11
2101	Pathogenicity and selective constraint on variation near splice sites. Genome Research, 2019, 29, 159-170.	2.4	70
2102	Reanalysis and optimisation of bioinformatic pipelines is critical for mutation detection. Human Mutation, 2019, 40, 374-379.	1.1	7
2103	A novel rare CUBN variant and three additional genes identified in Europeans with and without diabetes: results from an exome-wide association study of albuminuria. Diabetologia, 2019, 62, 292-305.	2.9	29
2104	Genetic analysis of neurodegenerative diseases in a pathology cohort. Neurobiology of Aging, 2019, 76, 214.e1-214.e9.	1.5	25
2105	Neurodevelopmental disease genes implicated by de novo mutation and copy number variation morbidity. Nature Genetics, 2019, 51, 106-116.	9.4	231
2106	An evolutionary framework for measuring epigenomic information and estimating cell-type-specific fitness consequences. Nature Genetics, 2019, 51, 335-342.	9.4	33
2107	Implications of de novo mutations in guiding drug discovery: A study of four neuropsychiatric disorders. Journal of Psychiatric Research, 2019, 110, 83-92.	1.5	3

#	Article	IF	Citations
2108	Rare variants in Protein tyrosine phosphatase, receptor type A (PTPRA) in schizophrenia: Evidence from a family based study. Schizophrenia Research, 2019, 206, 75-81.	1.1	6
2109	Blacklisting variants common in private cohorts but not in public databases optimizes human exome analysis. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 950-959.	3.3	52
2110	Widening the landscape of heritable pulmonary hypertension mutations in paediatric and adult cases. European Respiratory Journal, 2019, 53, 1801371.	3.1	72
2111	Whole-exome sequencing reveals SALL4 variants in premature ovarian insufficiency: an update on genotypeâ€“phenotype correlations. Human Genetics, 2019, 138, 83-92.	1.8	27
2112	Dysfunction of GRAP, encoding the GRB2-related adaptor protein, is linked to sensorineural hearing loss. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 1347-1352.	3.3	15
2113	CADD: predicting the deleteriousness of variants throughout the human genome. Nucleic Acids Research, 2019, 47, D886-D894.	6.5	2,360
2114	Human adenosine deaminase 2 deficiency: A multiâ€faceted inborn error of immunity. Immunological Reviews, 2019, 287, 62-72.	2.8	54
2115	Earlyâ€onset inflammatory bowel disease as a model disease to identify key regulators of immune homeostasis mechanisms. Immunological Reviews, 2019, 287, 162-185.	2.8	60
2116	Clinical whole exome sequencing in severe hypertriglyceridemia. Clinica Chimica Acta, 2019, 488, 31-39.	0.5	4
2117	Clinical and functional characterization of recurrent missense variants implicated inTHOC6-related intellectual disability. Human Molecular Genetics, 2019, 28, 952-960.	1.4	23
2118	Sphingolipid dysregulation due to lack of functional KDSR impairs proplatelet formation causing thrombocytopenia. Haematologica, 2019, 104, 1036-1045.	1.7	28
2119	The Burden of Candidate Pathogenic Variants for Kidney and Genitourinary Disorders Emerging From Exome Sequencing. Annals of Internal Medicine, 2019, 170, 11.	2.0	60
2120	A Multiplexed Assay for Exon Recognition Reveals that an Unappreciated Fraction of Rare Genetic Variants Cause Large-Effect Splicing Disruptions. Molecular Cell, 2019, 73, 183-194.e8.	4.5	88
2122	Homozygous PCDH12 variants result in phenotype of cerebellar ataxia, dystonia, retinopathy, and dysmorphism. Journal of Human Genetics, 2019, 64, 183-189.	1.1	13
2123	Functional Annotation of Animal Genomes (FAANG): Current Achievements and Roadmap. Annual Review of Animal Biosciences, 2019, 7, 65-88.	3.6	172
2124	Sequence variants in muscle tissueâ€related genes may determine the severity of muscle contractures in cerebral palsy. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2019, 180, 12-24.	1.1	4
2125	Complement C7is a novel risk gene for Alzheimer's disease in Han Chinese. National Science Review, 2019, 6, 257-274.	4.6	55
2126	Presence of tau astrogliopathy in frontotemporal dementia caused by a novel Grn nonsense (Trp2*) mutation. Neurobiology of Aging, 2019, 76, 214.e11-214.e15.	1.5	8

#	Article	IF	Citations
2127	Targeted panel sequencing in adult patients with left ventricular nonâ€compaction reveals a large genetic heterogeneity. Clinical Genetics, 2019, 95, 356-367.	1.0	56
2128	Severe hypertriglyceridemia is primarily polygenic. Journal of Clinical Lipidology, 2019, 13, 80-88.	0.6	136
2129	Elevation in Cell Cycle and Protein Metabolism Gene Transcription in Inactive Colonic Tissue From Icelandic Patients With Ulcerative Colitis. Inflammatory Bowel Diseases, 2019, 25, 317-327.	0.9	5
2130	Mutations in PLOD3, encoding lysyl hydroxylase 3, cause a complex connective tissue disorder including recessive dystrophic epidermolysis bullosa-like blistering phenotype with abnormal anchoring fibrils and type VII collagen deficiency. Matrix Biology, 2019, 81, 91-106.	1.5	45
2131	IL2RG hypomorphic mutation: identification of a novel pathogenic mutation in exon 8 and a review of the literature. Allergy, Asthma and Clinical Immunology, 2019, 15, 2.	0.9	31
2132	Identification of putative pathogenic single nucleotide variants (SNVs) in genes associated with heart disease in 290 cases of stillbirth. PLoS ONE, 2019, 14, e0210017.	1.1	24
2133	Recessive Rare Variants in Deoxyhypusine Synthase, an Enzyme Involved in the Synthesis of Hypusine, Are Associated with a Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 287-298.	2.6	38
2134	Identification of de novo CSNK2A1 and CSNK2B variants in cases of global developmental delay with seizures. Journal of Human Genetics, 2019, 64, 313-322.	1.1	51
2135	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	1.1	161
2136	Familial Sinus Node Disease Caused by a Gain of GIRK (G-Protein Activated Inwardly Rectifying K) Tj ETQq1 1 0.784314 rgBT /Overlock 1 e002238.	1.6	39
2137	Next generation sequencing and imprinting disorders: Current applications and future perspectives: Lessons from Silver-Russell syndrome. Molecular and Cellular Probes, 2019, 44, 1-7.	0.9	11
2138	Genome-wide meta-analysis identifies new loci and functional pathways influencing Alzheimerâ€™s disease risk. Nature Genetics, 2019, 51, 404-413.	9.4	1,625
2139	A guide to deep learning in healthcare. Nature Medicine, 2019, 25, 24-29.	15.2	1,906
2140	The Germline Variants rs61757955 and rs34988193 Are Predictive of Survival in Lower Grade Glioma Patients. Molecular Cancer Research, 2019, 17, 1075-1086.	1.5	7
2141	Efficient Variant Set Mixed Model Association Tests for Continuous and Binary Traits in Large-Scale Whole-Genome Sequencing Studies. American Journal of Human Genetics, 2019, 104, 260-274.	2.6	103
2142	Genome-Wide Association Study Meta-Analysis of the Alcohol Use Disorders Identification Test (AUDIT) in Two Population-Based Cohorts. American Journal of Psychiatry, 2019, 176, 107-118.	4.0	326
2143	Deep multi-region whole-genome sequencing reveals heterogeneity and gene-by-environment interactions in treatment-naive, metastatic lung cancer. Oncogene, 2019, 38, 1661-1675.	2.6	26
2144	Fine mapping of 14q13 reveals novel variants associated with different histological subtypes of papillary thyroid carcinoma. International Journal of Cancer, 2019, 144, 503-512.	2.3	4

#	Article	IF	Citations
2145	Burden of rare variants in causative genes for amyotrophic lateral sclerosis (ALS) accelerates age at onset of ALS. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 537-542.	0.9	28
2146	Detecting genetic modifiers of spondyloepimetaphyseal dysplasia with joint laxity in the Caucasian Afrikaner community. Human Molecular Genetics, 2019, 28, 1053-1063.	1.4	1
2147	Preliminary exploration of potential molecular therapeutic targets in recurrent and metastatic parathyroid carcinomas. International Journal of Cancer, 2019, 144, 525-532.	2.3	22
2148	Selective loss of function variants in IL6ST cause Hyper-IgE syndrome with distinct impairments of T-cell phenotype and function. Haematologica, 2019, 104, 609-621.	1.7	74
2149	Gene-specific metrics to facilitate identification of disease genes for molecular diagnosis in patient genomes: a systematic review. Briefings in Functional Genomics, 2019, 18, 23-29.	1.3	6
2150	Loss of function, missense, and intronic variants in NOTCH1 confer different risks for left ventricular outflow tract obstructive heart defects in two European cohorts. Genetic Epidemiology, 2019, 43, 215-226.	0.6	25
2151	Utility and implications of exome sequencing in earlyâ€onset Parkinson's disease. Movement Disorders, 2019, 34, 133-137.	2.2	36
2152	A map of constrained coding regions in the human genome. Nature Genetics, 2019, 51, 88-95.	9.4	201
2153	Multifactorial Inheritance and Complex Diseases. , 2019, , 323-358.		3
2154	Discovery of donor genotype associated with long-term survival of patients with hematopoietic stem cell transplantation in refractory acute myeloid leukemia. Leukemia and Lymphoma, 2019, 60, 1775-1781.	0.6	1
2155	Ensembl 2019. Nucleic Acids Research, 2019, 47, D745-D751.	6.5	879
2156	Identification of Candidate Genes for Mayer-Rokitansky-KÃ¼ster-Hauser Syndrome Using Genomic Approaches. Sexual Development, 2019, 13, 26-34.	1.1	29
2157	Next Generation Sequencing Analysis in Early Onset Dementia Patients. Journal of Alzheimer's Disease, 2019, 67, 243-256.	1.2	29
2158	Bioinformatics Tools in Clinical Genomics. , 2019, , 163-182.		0
2159	Discovery of common and rare genetic risk variants for colorectal cancer. Nature Genetics, 2019, 51, 76-87.	9.4	377
2160	Identification of CACNA1D variants associated with sinoatrial node dysfunction and deafness in additional Pakistani families reveals a clinical significance. Journal of Human Genetics, 2019, 64, 153-160.	1.1	32
2161	Global genetic insight contributed by consanguineous Pakistani families segregating hearing loss. Human Mutation, 2019, 40, 53-72.	1.1	48
2162	Predicting the Effect of Mutations on Protein Folding and Protein-Protein Interactions. Methods in Molecular Biology, 2019, 1851, 1-17.	0.4	12

#	Article	IF	Citations
2163	Analysis of Canadian multiple sclerosis patients does not support a role for FKBP6 in disease. Multiple Sclerosis Journal, 2019, 25, 1011-1013.	1.4	4
2164	Familial breast cancer and DNA repair genes: Insights into known and novel susceptibility genes from the GENESIS study, and implications for multigene panel testing. International Journal of Cancer, 2019, 144, 1962-1974.	2.3	50
2165	Molecular alterations associated with metastases of solid pseudopapillary neoplasms of the pancreas. Journal of Pathology, 2019, 247, 123-134.	2.1	32
2166	PLA2G6-associated neurodegeneration presenting as a complicated form of hereditary spastic paraplegia. Journal of Human Genetics, 2019, 64, 55-59.	1.1	17
2167	Genetic validation study of protein tyrosine phosphatase receptor type D (PTPRD) gene variants and risk for antipsychotic-induced weight gain. Journal of Neural Transmission, 2019, 126, 27-33.	1.4	13
2168	A review study: Computational techniques for expecting the impact of non-synonymous single nucleotide variants in human diseases. Gene, 2019, 680, 20-33.	1.0	47
2169	Machine learning for integrating data in biology and medicine: Principles, practice, and opportunities. Information Fusion, 2019, 50, 71-91.	11.7	340
2170	PRESM: personalized reference editor for somatic mutation discovery in cancer genomics. Bioinformatics, 2019, 35, 1445-1452.	1.8	6
2171	Phenotypic and genetic analysis of an adult cohort with extreme obesity. International Journal of Obesity, 2019, 43, 2057-2065.	1.6	5
2172	Estimating contribution of rare nonâ€coding variants to neuropsychiatric disorders. Psychiatry and Clinical Neurosciences, 2019, 73, 2-10.	1.0	18
2173	De novopattern discovery enables robust assessment of functional consequences of non-coding variants. Bioinformatics, 2019, 35, 1453-1460.	1.8	15
2174	Hypomorphic caspase activation and recruitment domain 11 (CARD11) mutations associated with diverse immunologic phenotypes with or without atopic disease. Journal of Allergy and Clinical Immunology, 2019, 143, 1482-1495.	1.5	116
2175	KAT6A Syndrome: genotypeâ€“phenotype correlation in 76 patients with pathogenic KAT6A variants. Genetics in Medicine, 2019, 21, 850-860.	1.1	68
2176	An optimized prediction framework to assess the functional impact of pharmacogenetic variants. Pharmacogenomics Journal, 2019, 19, 115-126.	0.9	109
2177	Integrating regulatory features data for prediction of functional disease-associated SNPs. Briefings in Bioinformatics, 2019, 20, 26-32.	3.2	11
2178	Understanding the disease genome: gene essentiality and the interplay of selection, recombination and mutation. Briefings in Bioinformatics, 2019, 20, 267-273.	3.2	11
2179	Frequently used bioinformatics tools overestimate the damaging effect of allelic variants. Genes and Immunity, 2019, 20, 10-22.	2.2	12
2180	Targeted deep sequencing of the PEAR1 locus for platelet aggregation in European and African American families. Platelets, 2019, 30, 380-386.	1.1	19

#	Article	IF	Citations
2181	Genetic variants and pathways implicated in a pediatric inflammatory bowel disease cohort. Genes and Immunity, 2019, 20, 131-142.	2.2	22
2182	Finding cancer driver mutations in the era of big data research. Biophysical Reviews, 2019, 11, 21-29.	1.5	20
2183	An integrative analysis of non-coding regulatory DNA variations associated with autism spectrum disorder. Molecular Psychiatry, 2019, 24, 1707-1719.	4.1	59
2184	Mutations in MERTK are not associated with age-related macular degeneration. International Ophthalmology, 2019, 39, 63-67.	0.6	2
2185	A combined analysis of genetically correlated traits identifies 187 loci and a role for neurogenesis and myelination in intelligence. Molecular Psychiatry, 2019, 24, 169-181.	4.1	238
2186	Genome-wide analysis reveals extensive genetic overlap between schizophrenia, bipolar disorder, and intelligence. Molecular Psychiatry, 2020, 25, 844-853.	4.1	156
2187	A joint study of whole exome sequencing and structural MRI analysis in major depressive disorder. Psychological Medicine, 2020, 50, 384-395.	2.7	19
2188	Genetic screening in early-onset Alzheimer's disease identified three novel presenilin mutations. Neurobiology of Aging, 2020, 86, 201.e9-201.e14.	1.5	16
2189	A novel mutation in PSEN1 (p.T119I) in an Argentine family with early- and late-onset Alzheimer's disease. Neurobiology of Aging, 2020, 85, 155.e9-155.e12.	1.5	15
2190	An extremely severe case of Aicardi-GoutiÃ¨res syndrome 7 with a novel variant in IFIH1. European Journal of Medical Genetics, 2020, 63, 103646.	0.7	12
2191	A genome-wide association study of tramadol metabolism from post-mortem samples. Pharmacogenomics Journal, 2020, 20, 94-103.	0.9	3
2192	Heterozygous rare genetic variants in non-syndromic early-onset obesity. International Journal of Obesity, 2020, 44, 830-841.	1.6	29
2193	Mutational hotspots of HSP47 and its potential role in cancer and bone-disorders. Genomics, 2020, 112, 552-566.	1.3	0
2194	Genetic contributions to two special factors of neuroticism are associated with affluence, higher intelligence, better health, and longer life. Molecular Psychiatry, 2020, 25, 3034-3052.	4.1	60
2195	Identification of novel loci associated with infant cognitive ability. Molecular Psychiatry, 2020, 25, 3010-3019.	4.1	6
2196	Diagnostic interpretation of genetic studies in patients with primary immunodeficiency diseases: AÂworking group report of the Primary Immunodeficiency Diseases Committee of the American Academy of Allergy, Asthma & Immunology. Journal of Allergy and Clinical Immunology, 2020, 145, 46-69.	1.5	54
2197	Novel HARS2 missense variants identified in individuals with sensorineural hearing impairment and Perrault syndrome. European Journal of Medical Genetics, 2020, 63, 103733.	0.7	9
2198	Genetic intolerance analysis as a tool for protein science. Biochimica Et Biophysica Acta - Biomembranes, 2020, 1862, 183058.	1.4	6

#	Article	IF	Citations
2199	Genes and variants in hematopoiesis-related pathways are associated with gemcitabine/carboplatin-induced thrombocytopenia. Pharmacogenomics Journal, 2020, 20, 179-191.	0.9	7
2200	Is there a duty to reinterpret genetic data? The ethical dimensions. Genetics in Medicine, 2020, 22, 633-639.	1.1	51
2201	Functional deficiency of DNA repair gene EXO5 results in androgen-induced genomic instability and prostate tumorigenesis. Oncogene, 2020, 39, 1246-1259.	2.6	8
2202	Novel features of PIK3CA-Related Overgrowth Spectrum: Lesson from an aborted fetus presenting a de novo constitutional PIK3CA mutation. European Journal of Medical Genetics, 2020, 63, 103775.	0.7	7
2203	Combined in vitro and in silico analyses of missense mutations in GNPTAB provide new insights into the molecular bases of mucolipidosis II and III alpha/beta. Human Mutation, 2020, 41, 133-139.	1.1	5
2204	Missense variants in TAF1 and developmental phenotypes: Challenges of determining pathogenicity. Human Mutation, 2020, 41, 449-464.	1.1	17
2205	EXOME REPORT: Novel mutation in ATP6V1B2 segregating with autosomal dominant epilepsy, intellectual disability and mild gingival and nail abnormalities. European Journal of Medical Genetics, 2020, 63, 103799.	0.7	14
2206	QTLbase: an integrative resource for quantitative trait loci across multiple human molecular phenotypes. Nucleic Acids Research, 2020, 48, D983-D991.	6.5	82
2207	Exome sequencing reveals a novel variant in NFX1 causing intracranial aneurysm in a Chinese family. Journal of NeuroInterventional Surgery, 2020, 12, 221-226.	2.0	7
2208	A genome-first approach to aggregating rare genetic variants in LMNA for association with electronic health record phenotypes. Genetics in Medicine, 2020, 22, 102-111.	1.1	42
2209	BRCA2 lossâ€ofâ€function germline mutations are associated with esophageal squamous cell carcinoma risk in Chinese. International Journal of Cancer, 2020, 146, 1042-1051.	2.3	26
2210	Frequent and Persistent PLCG1 Mutations in SÃ©zary Cells Directly Enhance PLCÎ³1 Activity and Stimulate NFÎºB, AP-1, and NFAT Signaling. Journal of Investigative Dermatology, 2020, 140, 380-389.e4.	0.3	25
2211	Predicting the effects of SNPs on transcription factor binding affinity. Bioinformatics, 2020, 36, 364-372.	1.8	37
2212	Diagnostic Approach to Monogenic Inflammatory Bowel Disease in Clinical Practice: A Ten-Year Multicentric Experience. Inflammatory Bowel Diseases, 2020, 26, 720-727.	0.9	32
2213	A clinical scoring system for congenital contractural arachnodactyly. Genetics in Medicine, 2020, 22, 124-131.	1.1	17
2214	Next generation sequencing panel in undifferentiated autoinflammatory diseases identifies patients with colchicine-responder recurrent fevers. Rheumatology, 2020, 59, 344-360.	0.9	36
2215	Identifying the Impact of Inframe Insertions and Deletions on Protein Function in Cancer. Journal of Computational Biology, 2020, 27, 786-795.	0.8	3
2216	Impact of natural neuromedinâ€B receptor variants on iron metabolism. American Journal of Hematology, 2020, 95, 167-177.	2.0	7

#	Article	IF	Citations
2217	A Pseudo-Blind Convolutional Neural Network for the Reduction of Compression Artifacts. IEEE Transactions on Circuits and Systems for Video Technology, 2020, 30, 1121-1135.	5.6	29
2218	The CYSMA web server: An example of integrative tool for in silico analysis of missense variants identified in Mendelian disorders. Human Mutation, 2020, 41, 375-386.	1.1	6
2219	Exome reports A de novo GNB2 variant associated with global developmental delay, intellectual disability, and dysmorphic features. European Journal of Medical Genetics, 2020, 63, 103804.	0.7	15
2220	Characterization of GDF2 Mutations and Levels of BMP9 and BMP10 in Pulmonary Arterial Hypertension. American Journal of Respiratory and Critical Care Medicine, 2020, 201, 575-585.	2.5	80
2221	Molecular profiles of BRCA1â€associated ovarian cancer treated by platinumâ€based therapy: Analysis of primary, residual and relapsed tumors. International Journal of Cancer, 2020, 146, 1879-1888.	2.3	18
2222	Prediction of impacts of mutations on protein structure and interactions: SDM, a statistical approach, and mCSM, using machine learning. Protein Science, 2020, 29, 247-257.	3.1	58
2223	A novel homozygous SACS mutation identified by whole exome sequencing-genotype phenotype correlations of all published cases. Journal of Molecular Neuroscience, 2020, 70, 131-141.	1.1	19
2224	A contemporary view on the molecular basis of neurodevelopmental disorders. , 2020, , 57-78.		0
2225	Matching whole genomes to rare genetic disorders: Identification of potential causative variants using phenotypeâ€weighted knowledge in the CAGI SickKids5 clinical genomes challenge. Human Mutation, 2020, 41, 347-362.	1.1	4
2226	Autosomal dominant Alport syndrome due to a COL4A4 mutation with an additional ESPN variant detected by whole-exome analysis. CEN Case Reports, 2020, 9, 59-64.	0.5	0
2227	Mutations in ASH1L confer susceptibility to Tourette syndrome. Molecular Psychiatry, 2020, 25, 476-490.	4.1	41
2228	An atypical case of KMT2Bâ€related dystonia manifesting asterixis and effect of deep brain stimulation of the globus pallidus. Neurology and Clinical Neuroscience, 2020, 8, 36-38.	0.2	4
2229	Genomeâ€wide association study of INDELs identified four novel susceptibility loci associated with lung cancer risk. International Journal of Cancer, 2020, 146, 2855-2864.	2.3	7
2230	Multiplex targeted highâ€throughput sequencing in a series of 352 patients with congenital limb malformations. Human Mutation, 2020, 41, 222-239.	1.1	17
2231	Genome-wide association and Mendelian randomisation analysis provide insights into the pathogenesis of heart failure. Nature Communications, 2020, 11, 163.	5.8	466
2232	Bioinformatics and Computational Tools for Next-Generation Sequencing Analysis in Clinical Genetics. Journal of Clinical Medicine, 2020, 9, 132.	1.0	126
2233	Shared Genetic Loci Between Body Mass Index and Major Psychiatric Disorders. JAMA Psychiatry, 2020, 77, 503.	6.0	82
2234	Targeted Next-Generation Sequencing in Patients with Suggestive X-Linked Intellectual Disability. Genes, 2020, 11, 51.	1.0	15

#	Article	IF	Citations
2235	Adaptor protein complex 4 deficiency: a paradigm of childhood-onset hereditary spastic paraplegia caused by defective protein trafficking. Human Molecular Genetics, 2020, 29, 320-334.	1.4	45
2236	Deep2Full: Evaluating strategies for selecting the minimal mutational experiments for optimal computational predictions of deep mutational scan outcomes. PLoS ONE, 2020, 15, e0227621.	1.1	8
2237	Pathogenicity Reclassification of RPE65 Missense Variants Related to Leber Congenital Amaurosis and Early-Onset Retinal Dystrophy. Genes, 2020, 11, 24.	1.0	14
2238	A novel de novo dominant mutation of NOTCH1 gene in an Iranian family with nonâ€syndromic congenital heart disease. Journal of Clinical Laboratory Analysis, 2020, 34, e23147.	0.9	13
2239	VPS13Dâ€related disorders presenting as a pure and complicated form of hereditary spastic paraplegia. Molecular Genetics & Genomic Medicine, 2020, 8, e1108.	0.6	29
2240	A novel heterozygous STAT5B variant in a patient with short stature and partial growth hormone insensitivity (GHI). Growth Hormone and IGF Research, 2020, 50, 61-70.	0.5	7
2241	Protein C deficiency; PROC gene variants in a Danish population. Thrombosis Research, 2020, 185, 153-159.	0.8	5
2242	Two unrelated pedigrees with achondrogenesis type 1b carrying a Japanâ€specific pathogenic variant in SLC26A2. American Journal of Medical Genetics, Part A, 2020, 182, 735-739.	0.7	0
2243	Prediction of mutation effects using a deep temporal convolutional network. Bioinformatics, 2020, 36, 2047-2052.	1.8	13
2244	A novel autosomal recessive lipodystrophy syndrome due to homozygous LMNA variant. Journal of Medical Genetics, 2020, 57, 422-426.	1.5	4
2245	Genetic variation in EPHA contributes to sensitivity to paclitaxelâ€induced peripheral neuropathy. British Journal of Clinical Pharmacology, 2020, 86, 880-890.	1.1	14
2246	The Melanocortin 4 Receptor p.Ile269Asn Mutation Is Associated with Childhood and Adult Obesity in Mexicans. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e1468-e1477.	1.8	9
2247	eQTL Analysis. Methods in Molecular Biology, 2020, , .	0.4	4
2248	Hypomorphic variants in AK2 reveal the contribution of mitochondrial function to B-cell activation. Journal of Allergy and Clinical Immunology, 2020, 146, 192-202.	1.5	13
2249	Exome sequencing for perinatal phenotypes: The significance of deep phenotyping. Prenatal Diagnosis, 2020, 40, 260-273.	1.1	20
2250	Prenatal whole exome sequencing detects a new homozygous fukutin (FKTN) mutation in a fetus with an ultrasound suspicion of familial Dandyâ€“Walker malformation. Molecular Genetics & Genomic Medicine, 2020, 8, e1054.	0.6	6
2251	Genetic Counseling and Genome Sequencing in Pediatric Rare Disease. Cold Spring Harbor Perspectives in Medicine, 2020, 10, a036632.	2.9	18
2252	Levetiracetam efficacy in PCDH19 Girls Clustering Epilepsy. European Journal of Paediatric Neurology, 2020, 24, 142-147.	0.7	16

#	Article	IF	Citations
2253	Regulatory genome variants in human susceptibility to infection. Human Genetics, 2020, 139, 759-768.	1.8	14
2254	Multiple rare inherited variants in a four generation schizophrenia family offer leads for complex mode of disease inheritance. Schizophrenia Research, 2020, 216, 288-294.	1.1	10
2255	Identification of novel FBN1 variations implicated in congenital scoliosis. Journal of Human Genetics, 2020, 65, 221-230.	1.1	20
2256	Identification of a homozygous VRK1 mutation in two patients with adultâ€onset distal hereditary motor neuropathy. Muscle and Nerve, 2020, 61, 395-400.	1.0	10
2257	Ribosomal protein gene RPL9 variants can differentially impair ribosome function and cellular metabolism. Nucleic Acids Research, 2020, 48, 770-787.	6.5	28
2258	Exome sequencing identifies de novo splicing variant in XRCC6 in sporadic case of autism. Journal of Human Genetics, 2020, 65, 287-296.	1.1	8
2259	Next-Generation Sequencing Identifies BRCA1 and/or BRCA2 Mutations in Women at High Hereditary Risk for Breast Cancer with Shorter Telomere Length. OMICS A Journal of Integrative Biology, 2020, 24, 5-15.	1.0	4
2260	Systematic analysis of alterations in the ubiquitin proteolysis system reveals its contribution to driver mutations in cancer. Nature Cancer, 2020, 1, 122-135.	5.7	30
2261	Highâ€throughput custom capture sequencing identifies novel mutations in colobomaâ€associated genes: Mutation in DNAâ€binding domain of retinoic acid receptor beta affects nuclear localization causing ocular coloboma. Human Mutation, 2020, 41, 678-695.	1.1	13
2262	Insights into the genetic basis of retinal detachment. Human Molecular Genetics, 2020, 29, 689-702.	1.4	26
2263	A birdâ€™s-eye view of Italian genomic variation through whole-genome sequencing. European Journal of Human Genetics, 2020, 28, 435-444.	1.4	29
2264	De novo CLTC variants are associated with a variable phenotype from mild to severe intellectual disability, microcephaly, hypoplasia of the corpus callosum, and epilepsy. Genetics in Medicine, 2020, 22, 797-802.	1.1	15
2265	Punctiform and Polychromatic Pre-Descemet Corneal Dystrophy: Clinical Evaluation and Identification of the Genetic Basis. American Journal of Ophthalmology, 2020, 212, 88-97.	1.7	6
2266	Biallelic Mutations in MTPAP Associated with a Lethal Encephalopathy. Neuropediatrics, 2020, 51, 178-184.	0.3	3
2267	Identification of novel mutations of ovarian cancer-related genes from RNA-sequencing data for Japanese epithelial ovarian cancer patients. Endocrine Journal, 2020, 67, 219-229.	0.7	6
2268	Glycogen Storage Disease Type IV Diagnosed at Fetal Autopsy. Pediatric and Developmental Pathology, 2020, 23, 301-305.	0.5	3
2269	Targeted exon sequencing in deceased schizophrenia patients in Denmark. International Journal of Legal Medicine, 2020, 134, 135-147.	1.2	2
2270	Two pathologically confirmed cases of novel mutations in the MAPT gene causing frontotemporal dementia. Neurobiology of Aging, 2020, 87, 141.e15-141.e20.	1.5	3

#	Article	IF	Citations
2271	Genetic polymorphisms in collagen-related genes are associated with pelvic organ prolapse. Menopause, 2020, 27, 223-229.	0.8	17
2272	Identification of pathogenic variant enriched regions across genes and gene families. Genome Research, 2020, 30, 62-71.	2.4	47
2273	Expanding Clinical Phenotype and Novel Insights into the Pathogenesis of ICOS Deficiency. Journal of Clinical Immunology, 2020, 40, 277-288.	2.0	21
2274	InMeRF: prediction of pathogenicity of missense variants by individual modeling for each amino acid substitution. NAR Genomics and Bioinformatics, 2020, 2, lqaa038.	1.5	16
2275	A whole-genome sequenced control population in northern Sweden reveals subregional genetic differences. PLoS ONE, 2020, 15, e0237721.	1.1	1
2276	An All-In-One Transcriptome-Based Assay to Identify Therapy-Guiding Genomic Aberrations in Nonsmall Cell Lung Cancer Patients. Cancers, 2020, 12, 2843.	1.7	6
2277	Ovarian Cancer Risk Variants Are Enriched in Histotype-Specific Enhancers and Disrupt Transcription Factor Binding Sites. American Journal of Human Genetics, 2020, 107, 622-635.	2.6	14
2278	Base editing: advances and therapeutic opportunities. Nature Reviews Drug Discovery, 2020, 19, 839-859.	21.5	218
2279	Pathogenesis of CDK8-associated disorder: two patients with novel CDK8 variants and in vitro and in vivo functional analyses of the variants. Scientific Reports, 2020, 10, 17575.	1.6	7
2280	Ultrafast and scalable variant annotation and prioritization with big functional genomics data. Genome Research, 2020, 30, 1789-1801.	2.4	14
2281	Use of patient derived urine renal epithelial cells to confirm pathogenicity of PKHD1 alleles. BMC Nephrology, 2020, 21, 435.	0.8	13
2282	Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia. Brain, 2020, 143, 2929-2944.	3.7	29
2283	Neurological disorder-associated genetic variants in individuals with psychogenic nonepileptic seizures. Scientific Reports, 2020, 10, 15205.	1.6	12
2284	Genomic Profiling Identified ERCC2 E606Q Mutation in Helicase Domain Respond to Platinum-Based Neoadjuvant Therapy in Urothelial Bladder Cancer. Frontiers in Oncology, 2020, 10, 1643.	1.3	4
2285	Mutations disrupting neuritogenesis genes confer risk for cerebral palsy. Nature Genetics, 2020, 52, 1046-1056.	9.4	96
2286	Biological insights from multi-omic analysis of 31 genomic risk loci for adult hearing difficulty. PLoS Genetics, 2020, 16, e1009025.	1.5	42
2287	The shaping of immunological responses through natural selection after the Roma Diaspora. Scientific Reports, 2020, 10, 16134.	1.6	2
2288	Genome-Wide Association Study of Suicide Death and Polygenic Prediction of Clinical Antecedents. American Journal of Psychiatry, 2020, 177, 917-927.	4.0	66

#	Article	IF	Citations
2289	Exome Sequencing Implicates Impaired GABA Signaling and Neuronal Ion Transport in Trigeminal Neuralgia. IScience, 2020, 23, 101552.	1.9	32
2290	Neurogenetic fetal akinesia and arthrogryposis: genetics, expanding genotype-phenotypes and functional genomics. Journal of Medical Genetics, 2021, 58, 609-618.	1.5	46
2291	A Novel CCT5 Missense Variant Associated with Early Onset Motor Neuropathy. International Journal of Molecular Sciences, 2020, 21, 7631.	1.8	8
2292	Monogenic variants in dystonia: an exome-wide sequencing study. Lancet Neurology, The, 2020, 19, 908-918.	4.9	139
2293	Cancer Predisposition Genes in Cancer-Free Families. Cancers, 2020, 12, 2770.	1.7	2
2294	Prioritizing sequence variants in conserved non-coding elements in the chicken genome using chCADD. PLoS Genetics, 2020, 16, e1009027.	1.5	7
2295	A Recurrent Pathogenic Variant of INPP5K Underlies Autosomal Recessive Congenital Muscular Dystrophy With Cataracts and Intellectual Disability: Evidence for a Founder Effect in Southern Italy. Frontiers in Genetics, 2020, 11, 565868.	1.1	8
2296	Novel loci for childhood body mass index and shared heritability with adult cardiometabolic traits. PLoS Genetics, 2020, 16, e1008718.	1.5	95
2297	Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature, 2020, 586, 757-762.	13.7	343
2298	Using Co-segregation and Loss of Heterozygosity Analysis to Define the Pathogenicity of Unclassified Variants in Hereditary Breast Cancer Patients. Frontiers in Oncology, 2020, 10, 571330.	1.3	5
2299	Essential role of autophagy in restricting poliovirus infection revealed by identification of an ATG7 defect in a poliomyelitis patient. Autophagy, 2021, 17, 2449-2464.	4.3	10
2300	CRB1-related retinopathy overlapping the ocular phenotype of S-adenosylhomocysteine hydrolase deficiency. Ophthalmic Genetics, 2020, 41, 457-464.	0.5	3
2301	Overrepresentation of genetic variation in the AnkyrinG interactome is related to a range of neurodevelopmental disorders. European Journal of Human Genetics, 2020, 28, 1726-1733.	1.4	4
2302	PWAS: proteome-wide association studyâ€”linking genes and phenotypes by functional variation in proteins. Genome Biology, 2020, 21, 173.	3.8	52
2303	Unified inference of missense variant effects and gene constraints in the human genome. PLoS Genetics, 2020, 16, e1008922.	1.5	17
2304	Genetic association of gemcitabine/carboplatin-induced leukopenia and neutropenia in non-small cell lung cancer patients using whole-exome sequencing. Lung Cancer, 2020, 147, 106-114.	0.9	5
2305	Whole exome sequencing identified mutations causing hearing loss in five consanguineous Pakistani families. BMC Medical Genetics, 2020, 21, 151.	2.1	8
2306	An activating germline IDH1 variant associated with a tumor entity characterized by unilateral and bilateral chondrosarcoma of the mastoid. Human Genetics and Genomics Advances, 2020, 1, 100006.	1.0	3

#	Article	IF	Citations
2307	SVFX: a machine learning framework to quantify the pathogenicity of structural variants. Genome Biology, 2020, 21, 274.	3.8	24
2308	Role of Non-Coding Variants in Brugada Syndrome. International Journal of Molecular Sciences, 2020, 21, 8556.	1.8	6
2309	Precise uncertain significance prediction using latent space matrix factorization models: genomics variant and heterogeneous clinical data-driven approaches. Briefings in Bioinformatics, 2021, 22, .	3.2	4
2310	Characterization of an N-terminal Nav1.5 channel variant â€“ a potential risk factor for arrhythmias and sudden death?. BMC Medical Genetics, 2020, 21, 227.	2.1	1
2311	Inferring the molecular and phenotypic impact of amino acid variants with MutPred2. Nature Communications, 2020, 11, 5918.	5.8	305
2312	Discovery of rare variants associated with blood pressure regulation through meta-analysis of 1.3 million individuals. Nature Genetics, 2020, 52, 1314-1332.	9.4	91
2313	Monoallelic Mutations in CC2D1A Suggest a Novel Role in Human Heterotaxy and Ciliary Dysfunction. Circulation Genomic and Precision Medicine, 2020, 13, e003000.	1.6	4
2314	Lack of pathogenic germline DICER1 variants in males with testicular germ-cell tumors. Cancer Genetics, 2020, 248-249, 49-56.	0.2	0
2315	The genetic architecture of sporadic and multiple consecutive miscarriage. Nature Communications, 2020, 11, 5980.	5.8	52
2316	Candidate Gene Discovery in Hereditary Colorectal Cancer and Polyposis Syndromesâ€“Considerations for Future Studies. International Journal of Molecular Sciences, 2020, 21, 8757.	1.8	7
2317	Currently Applied Molecular Assays for Identifying ESR1 Mutations in Patients with Advanced Breast Cancer. International Journal of Molecular Sciences, 2020, 21, 8807.	1.8	6
2318	FI-Net: Identification of Cancer Driver Genes by Using Functional Impact Prediction Neural Network. Frontiers in Genetics, 2020, 11, 564839.	1.1	8
2319	Detecting Genetic Ancestry and Adaptation in the Taiwanese Han People. Molecular Biology and Evolution, 2021, 38, 4149-4165.	3.5	12
2320	â€˜There and Back Againâ€™â€”Forward Genetics and Reverse Phenotyping in Pulmonary Arterial Hypertension. Genes, 2020, 11, 1408.	1.0	11
2321	Systematic Review of Sequencing Studies and Gene Expression Profiling in Familial Meniere Disease. Genes, 2020, 11, 1414.	1.0	15
2322	Common germline-somatic variant interactions in advanced urothelial cancer. Nature Communications, 2020, 11, 6195.	5.8	21
2323	A comparative genomics multitool for scientific discovery and conservation. Nature, 2020, 587, 240-245.	13.7	216
2324	Prediction of driver variants in the cancer genome via machine learning methodologies. Briefings in Bioinformatics, 2021, 22, .	3.2	13

#	Article	IF	Citations
2325	Dataset on amelogenesis-related genes variants (ENAM and ENAM interacting genes) and on human leukocyte antigen alleles (DQ2 and DQ8) distribution in children with and without molar-incisor hypomineralisation (MIH). Data in Brief, 2020, 32, 106224.	0.5	2
2326	RRP7A links primary microcephaly to dysfunction of ribosome biogenesis, resorption of primary cilia, and neurogenesis. Nature Communications, 2020, 11, 5816.	5.8	34
2327	CD70 Deficiency Associated With Chronic Epstein-Barr Virus Infection, Recurrent Airway Infections and Severe Gingivitis in a 24-Year-Old Woman. Frontiers in Immunology, 2020, 11, 1593.	2.2	3
2328	Pathogenic and likely pathogenic variants in at least five genes account for approximately 3% of mild isolated nonsyndromic thrombocytopenia. Transfusion, 2020, 60, 2419-2431.	0.8	6
2329	The possible influence of genetic aetiological factors on molarâ€“incisor hypomineralisation. Archives of Oral Biology, 2020, 118, 104848.	0.8	23
2330	VEPAD - Predicting the effect of variants associated with Alzheimer's disease using machine learning. Computers in Biology and Medicine, 2020, 124, 103933.	3.9	14
2331	Genetic correlation, pleiotropy, and causal associations between substance use and psychiatric disorder. Psychological Medicine, 2022, 52, 968-978.	2.7	41
2332	The role of exome sequencing in newborn screening for inborn errors of metabolism. Nature Medicine, 2020, 26, 1392-1397.	15.2	112
2333	Genetics and major depressive disorder: clinical implications for disease risk, prognosis and treatment. International Clinical Psychopharmacology, 2020, 35, 233-242.	0.9	22
2334	Epigenomic and Transcriptomic Dynamics During Human Heart Organogenesis. Circulation Research, 2020, 127, e184-e209.	2.0	27
2335	Informing patients about their mutation tests: CDKN2A c.256G>A in melanoma as an example. Hereditary Cancer in Clinical Practice, 2020, 18, 15.	0.6	3
2336	Identification of genetic loci jointly influencing coronary artery disease risk and sleep traits of insomnia, sleep duration, and chronotype. Sleep Medicine, 2020, 74, 116-123.	0.8	6
2337	Genetic variation in Charcotâ€“Marieâ€“Tooth genes contributes to sensitivity to paclitaxel-induced peripheral neuropathy. Pharmacogenomics, 2020, 21, 841-851.	0.6	7
2338	Clinical, neuropathological, and genetic characterization of STUB1 variants in cerebellar ataxias: a frequent cause of predominant cognitive impairment. Genetics in Medicine, 2020, 22, 1851-1862.	1.1	30
2339	The genetic architecture of human brainstem structures and their involvement in common brain disorders. Nature Communications, 2020, 11, 4016.	5.8	26
2340	Gene-wise variant burden and genomic characterization of nearly every gene. Pharmacogenomics, 2020, 21, 827-840.	0.6	2
2341	Differences in local population history at the finest level: the case of the Estonian population. European Journal of Human Genetics, 2020, 28, 1580-1591.	1.4	23
2342	Novel manifestations of immune dysregulation and granule defects in gray platelet syndrome. Blood, 2020, 136, 1956-1967.	0.6	34

#	Article	IF	Citations
2343	Deep sequencing shows that accumulation of potentially pathogenic mtDNA mutations rather than mtDNA copy numbers may be associated with early embryonic loss. Journal of Assisted Reproduction and Genetics, 2020, 37, 2181-2188.	1.2	1
2344	An NGS-based genotyping in LQTS; minor genes are no longer minor. Journal of Human Genetics, 2020, 65, 1083-1091.	1.1	10
2345	Integration of GWAS and eQTL Analysis to Identify Risk Loci and Susceptibility Genes for Gastric Cancer. Frontiers in Genetics, 2020, 11, 679.	1.1	10
2346	Exome Sequencing Reveals Signal Transduction Genes Involved in Impulse Control Disorders in Parkinson's Disease. Frontiers in Neurology, 2020, 11, 641.	1.1	3
2347	Usher syndrome and Nebulinâ€associated myopathy in a single patient due to variants in MYO7A and NEB. Clinical Case Reports (discontinued), 2020, 8, 2476-2482.	0.2	0
2348	Gene mapping and functional annotation of GWAS of oral ulcers using FUMA software. Scientific Reports, 2020, 10, 12205.	1.6	6
2349	Functional fine-mapping of noncoding risk variants in amyotrophic lateral sclerosis utilizing convolutional neural network. Scientific Reports, 2020, 10, 12872.	1.6	11
2350	RADAR: annotation and prioritization of variants in the post-transcriptional regulome of RNA-binding proteins. Genome Biology, 2020, 21, 151.	3.8	9
2351	Prenatal clinical manifestations in individuals with COL4A1/2 variants. Journal of Medical Genetics, 2021, 58, 505-513.	1.5	22
2352	MISTIC: A prediction tool to reveal disease-relevant deleterious missense variants. PLoS ONE, 2020, 15, e0236962.	1.1	26
2353	SLC38A8 mutations result in arrested retinal development with loss of cone photoreceptor specialization. Human Molecular Genetics, 2020, 29, 2989-3002.	1.4	28
2354	SLC12A2 mutations cause NKCC1 deficiency with encephalopathy and impaired secretory epithelia. Neurology: Genetics, 2020, 6, e478.	0.9	20
2355	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. Brain, 2020, 143, 2437-2453.	3.7	21
2356	parSMURF, a high-performance computing tool for the genome-wide detection of pathogenic variants. GigaScience, 2020, 9, .	3.3	11
2357	Otitis media susceptibility and shifts in the head and neck microbiome due to SPINK5 variants. Journal of Medical Genetics, 2021, 58, 442-452.	1.5	14
2358	The utility of whole exome sequencing for identification of the molecular etiology in autosomal recessive developmental and epileptic encephalopathies. Neurological Sciences, 2020, 41, 3729-3739.	0.9	5
2359	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	2.6	48
2360	Neanderthal introgression reintroduced functional ancestral alleles lost in Eurasian populations. Nature Ecology and Evolution, 2020, 4, 1332-1341.	3.4	33

#	Article	IF	Citations
2361	Genetics of extreme human longevity to guide drug discovery for healthy ageing. Nature Metabolism, 2020, 2, 663-672.	5.1	32
2362	Predicting functional effects of missense variants in voltage-gated sodium and calcium channels. Science Translational Medicine, 2020, 12, .	5.8	84
2363	A Novel Syndrome With Short Stature, Mandibular Hypoplasia, and Osteoporosis May Be Associated With a PRRT3 Variant. Journal of the Endocrine Society, 2020, 4, bvaa088.	0.1	0
2364	Genetic background of ataxia in children younger than 5 years in Finland. Neurology: Genetics, 2020, 6, e444.	0.9	6
2365	Loss of CBY1 results in a ciliopathy characterized by features of Joubert syndrome. Human Mutation, 2020, 41, 2179-2194.	1.1	16
2366	Comprehensive characterization of amino acid positions in protein structures reveals molecular effect of missense variants. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 28201-28211.	3.3	68
2367	Two Compound Heterozygous Variants in SNX14 Cause Stereotypies and Dystonia in Autosomal Recessive Spinocerebellar Ataxia 20. Frontiers in Genetics, 2020, 11, 1038.	1.1	2
2368	Two Variants in the NOTCH4 and HLA-C Genes Contribute to Familial Clustering of Psoriasis. International Journal of Genomics, 2020, 2020, 1-9.	0.8	2
2369	The First Report of Biallelic Missense Mutations in the SFRP4 Gene Causing Pyle Disease in Two Siblings. Frontiers in Genetics, 2020, 11, 593407.	1.1	8
2370	Multi-omic studies on missense PLG variants in families with otitis media. Scientific Reports, 2020, 10, 15035.	1.6	4
2371	Identification of potential causal variants for premature ovarian failure by whole exome sequencing. BMC Medical Genomics, 2020, 13, 159.	0.7	12
2372	Two New Cases of Hypertrophic Cardiomyopathy and Skeletal Muscle Features Associated with ALPK3 Homozygous and Compound Heterozygous Variants. Genes, 2020, 11, 1201.	1.0	20
2373	Identification of a functional SNP rs7304782 at schizophrenia risk locus 12q24.31 and validation of its association with schiz ophrenia in Chinese populations. Psychiatry Research, 2020, 294, 113491.	1.7	1
2374	Functional analysis of BRCA1 RING domain variants: computationally derived structural data can improve upon experimental features for training predictive models. Integrative Biology (United) Tj ETQq1 1 0.784314 rgBT /Overlock 10		
2375	Lights and Shadows in the Genetics of Syndromic and Non-Syndromic Hearing Loss in the Italian Population. Genes, 2020, 11, 1237.	1.0	13
2376	Dominant-negative mutations in human IL6ST underlie hyper-IgE syndrome. Journal of Experimental Medicine, 2020, 217, .	4.2	64
2377	Genome sequencing unveils mutational landscape of the familial Mediterranean fever: Potential implications of IL33/ST2 signalling. Journal of Cellular and Molecular Medicine, 2020, 24, 11294-11306.	1.6	7
2378	Systems genetics analysis identifies calcium-signaling defects as novel cause of congenital heart disease. Genome Medicine, 2020, 12, 76.	3.6	15

#	Article	IF	Citations
2379	Genome-Wide Search for SNP Interactions in GWAS Data: Algorithm, Feasibility, Replication Using Schizophrenia Datasets. Frontiers in Genetics, 2020, 11, 1003.	1.1	14
2381	Molecular testing for the study of non-syndromic hearing loss. Hearing, Balance and Communication, 2020, 18, 270-277.	0.1	5
2382	Dominant mutations in ITPR3 cause Charcotâ€Marieâ€Tooth disease. Annals of Clinical and Translational Neurology, 2020, 7, 1962-1972.	1.7	9
2383	Variants of genes encoding TNF receptors and ligands and proteins regulating TNF activation in familial multiple sclerosis. CNS Neuroscience and Therapeutics, 2020, 26, 1178-1184.	1.9	4
2384	Characterization of Two Novel Variants of the Steroidogenic Acute Regulatory Protein Identified in a Girl with Classic Lipoid Congenital Adrenal Hyperplasia. International Journal of Molecular Sciences, 2020, 21, 6185.	1.8	3
2385	Two Novel Pathogenic Variants Confirm RMND1 Causative Role in Perrault Syndrome with Renal Involvement. Genes, 2020, 11, 1060.	1.0	16
2386	A Membrane-Tethered Ubiquitination Pathway Regulates Hedgehog Signaling and Heart Development. Developmental Cell, 2020, 55, 432-449.e12.	3.1	21
2387	A Novel Recurrent COL5A1 Genetic Variant Is Associated With a Dysplasia-Associated Arterial Disease Exhibiting Dissections and Fibromuscular Dysplasia. Arteriosclerosis, Thrombosis, and Vascular Biology, 2020, 40, 2686-2699.	1.1	30
2388	Annotation of Human Exome Gene Variants with Consensus Pathogenicity. Genes, 2020, 11, 1076.	1.0	4
2389	Retrospective evaluation of whole exome and genome mutation calls in 746 cancer samples. Nature Communications, 2020, 11, 4748.	5.8	27
2390	CAPICE: a computational method for Consequence-Agnostic Pathogenicity Interpretation of Clinical Exome variations. Genome Medicine, 2020, 12, 75.	3.6	30
2391	Whole exome sequencing revealed a novel homozygous variant in the DGKE catalytic domain: a case report of familial hemolytic uremic syndrome. BMC Medical Genetics, 2020, 21, 169.	2.1	2
2392	Novel Mutation C.7348C>T in NF1 Gene Identified by Whole-Exome Sequencing in Patient with Overlapping Clinical Symptoms of Neurofibromatosis Type 1 and Bannayanâ€“Rileyâ€“Ruvalcaba Syndrome. Cytology and Genetics, 2020, 54, 353-362.	0.2	0
2393	PCDH19 Pathogenic Variants in Males: Expanding the Phenotypic Spectrum. Advances in Experimental Medicine and Biology, 2020, 1298, 177-187.	0.8	15
2394	Rapid progressive ALS in a patient with a DNAJC7 loss-of-function mutation. Neurology: Genetics, 2020, 6, e503.	0.9	13
2395	Assessing performance of pathogenicity predictors using clinically relevant variant datasets. Journal of Medical Genetics, 2021, 58, 547-555.	1.5	57
2396	Efficient geneâ€“environment interaction tests for large biobankâ€scale sequencing studies. Genetic Epidemiology, 2020, 44, 908-923.	0.6	15
2397	Mutational burden and potential oligogenic model of TBX6â€mediated genes in congenital scoliosis. Molecular Genetics & Genomic Medicine, 2020, 8, e1453.	0.6	6

#	Article	IF	Citations
2398	Genetic testing in Poland and Ukraine: should comprehensive germline testing of BRCA1 and BRCA2 be recommended for women with breast and ovarian cancer?. Genetical Research, 2020, 102, e6.	0.3	12
2399	Transcriptomic signatures across human tissues identify functional rare genetic variation. Science, 2020, 369, .	6.0	89
2400	A Deep Learning Framework Identifies Pathogenic Noncoding Somatic Mutations from Personal Prostate Cancer Genomes. Cancer Research, 2020, 80, 4644-4654.	0.4	9
2401	A polyclonal allelic expression assay for detecting regulatory effects of transcript variants. Genome Medicine, 2020, 12, 79.	3.6	5
2402	Novel CHP1 mutation in autosomal-recessive cerebellar ataxia: autopsy features of two siblings. Acta Neuropathologica Communications, 2020, 8, 134.	2.4	1
2403	SVAD: A genetic database curates non-ischemic sudden cardiac death-associated variants. PLoS ONE, 2020, 15, e0237731.	1.1	0
2404	Spectrum of clinical features and genetic variants in mevalonate kinase (MVK) gene of South Indian families suffering from Hyperimmunoglobulin D Syndrome. PLoS ONE, 2020, 15, e0237999.	1.1	12
2405	Selective targeting of cancer signaling pathways with nanomedicines: challenges and progress. Future Oncology, 2020, 16, 2959-2979.	1.1	22
2406	A novel homozygous variant in REN in a family presenting with classic features of disorders involving the reninâ€“angiotensin pathway, without renal tubular dysgenesis. American Journal of Medical Genetics, Part A, 2020, 182, 2284-2290.	0.7	3
2407	Dynamic incorporation of multiple in silico functional annotations empowers rare variant association analysis of large whole-genome sequencing studies at scale. Nature Genetics, 2020, 52, 969-983.	9.4	146
2408	A method for scoring the cell type-specific impacts of noncoding variants in personal genomes. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 21364-21372.	3.3	16
2409	The Needle in the Haystackâ€”Searching for Genetic and Epigenetic Differences in Monozygotic Twins Discordant for Tetralogy of Fallot. Journal of Cardiovascular Development and Disease, 2020, 7, 55.	0.8	10
2410	Identification and characterization of constrained non-exonic bases lacking predictive epigenomic and transcription factor binding annotations. Nature Communications, 2020, 11, 6168.	5.8	1
2411	Improving the informativeness of Mendelian disease-derived pathogenicity scores for common disease. Nature Communications, 2020, 11, 6258.	5.8	8
2412	A flexible computational pipeline for research analyses of unsolved clinical exome cases. Npj Genomic Medicine, 2020, 5, 54.	1.7	4
2413	Mutational analysis in familial Alzheimerâ€™s disease of Han Chinese in Taiwan with a predominant mutation PSEN1 p.Met146Ile. Scientific Reports, 2020, 10, 19769.	1.6	7
2414	Mutation Frequency in Main Susceptibility Genes Among Patients With Head and Neck Paragangliomas. Frontiers in Genetics, 2020, 11, 614908.	1.1	16
2415	A Wide Spectrum of Genetic Disorders Causing Severe Childhood Epilepsy in Taiwan: A Case Series of Ultrarare Genetic Cause and Novel Mutation Analysis in a Pilot Study. Journal of Personalized Medicine, 2020, 10, 281.	1.1	15

#	Article	IF	Citations
2416	Genetic and Clinical Profiles of Pheochromocytoma and Paraganglioma: A Single Center Study. Frontiers in Endocrinology, 2020, 11, 574662.	1.5	14
2417	Cell Type-Specific Annotation and Fine Mapping of Variants Associated With Brain Disorders. Frontiers in Genetics, 2020, 11, 575928.	1.1	2
2418	Parkinsonâ€™s Disease in Teneurin Transmembrane Protein 4 (TENM4) Mutation Carriers. Frontiers in Genetics, 2020, 11, 598064.	1.1	6
2419	Clustering Analysis Supports the Detection of Biological Processes Related to Autism Spectrum Disorder. Genes, 2020, 11, 1476.	1.0	8
2420	Prioritizing long range interactions in noncoding regions using GWAS and deletions perturbed TADs. Computational and Structural Biotechnology Journal, 2020, 18, 2945-2952.	1.9	2
2421	MosaicBase: A Knowledgebase of Postzygotic Mosaic Variants in Noncancer Disease-related and Healthy Human Individuals. Genomics, Proteomics and Bioinformatics, 2020, 18, 140-149.	3.0	10
2422	Multiplexed Functional Assessment of Genetic Variants in CARD11. American Journal of Human Genetics, 2020, 107, 1029-1043.	2.6	38
2423	Human T-bet Governs Innate and Innate-like Adaptive IFN-Î³ Immunity against Mycobacteria. Cell, 2020, 183, 1826-1847.e31.	13.5	83
2424	Computational methods for detecting cancer hotspots. Computational and Structural Biotechnology Journal, 2020, 18, 3567-3576.	1.9	12
2425	Whole-genome sequencing analysis of the cardiometabolic proteome. Nature Communications, 2020, 11, 6336.	5.8	38
2426	Improving existing analysis pipeline to identify and analyze cancer driver genes using multi-omics data. Scientific Reports, 2020, 10, 20521.	1.6	10
2427	NGS Panel Testing of Triple-Negative Breast Cancer Patients in Cyprus: A Study of BRCA-Negative Cases. Cancers, 2020, 12, 3140.	1.7	8
2428	Genome-wide meta-analysis of brain volume identifies genomic loci and genes shared with intelligence. Nature Communications, 2020, 11, 5606.	5.8	50
2429	Unsupervised Clustering of Missense Variants in HNF1A Using Multidimensional Functional Data Aids Clinical Interpretation. American Journal of Human Genetics, 2020, 107, 670-682.	2.6	25
2430	Whole-exome sequencing in patients with protein aggregate myopathies reveals causative mutations associated with novel atypical phenotypes. Neurological Sciences, 2021, 42, 2819-2827.	0.9	2
2431	The genetic landscape of axonal neuropathies in the middle-aged and elderly. Neurology, 2020, 95, e3163-e3179.	1.5	19
2432	De novo mutations in TOMM70, a receptor of the mitochondrial import translocase, cause neurological impairment. Human Molecular Genetics, 2020, 29, 1568-1579.	1.4	29
2433	Variant discovery using next-generation sequencing and its future role in pharmacogenetics. Pharmacogenomics, 2020, 21, 471-486.	0.6	9

#	Article	IF	Citations
2434	Germline whole exome sequencing of a family with appendiceal mucinous tumours presenting with pseudomyxoma peritonei. BMC Cancer, 2020, 20, 369.	1.1	5
2435	Clinical and Genetic Characterization of Autosomal Recessive Spinocerebellar Ataxia Type 16 (SCAR16) in Taiwan. Cerebellum, 2020, 19, 544-549.	1.4	13
2436	Systems Analysis Implicates WAVE2ÂComplex in the Pathogenesis ofÂDevelopmental Left-Sided ObstructiveÂHeart Defects. JACC Basic To Translational Science, 2020, 5, 376-386.	1.9	15
2437	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
2438	Tropomyosin 1 genetically constrains in vitro hematopoiesis. BMC Biology, 2020, 18, 52.	1.7	8
2439	Family-based genome-wide association study of leprosy in Vietnam. PLoS Pathogens, 2020, 16, e1008565.	2.1	8
2440	A Novel LINS1 Truncating Mutation in Autosomal Recessive Nonsyndromic Intellectual Disability. Frontiers in Psychiatry, 2020, 11, 354.	1.3	2
2441	Identification of <i>Transient Receptor Potential Channel 4-Associated Protein</i> as a Novel Candidate Gene Causing Congenital Primary Hypothyroidism. Hormone Research in Paediatrics, 2020, 93, 16-29.	0.8	7
2442	Mutations in MC4R facilitate the angiogenic activity in patients with orbital venous malformation. Experimental Biology and Medicine, 2020, 245, 956-963.	1.1	0
2443	Enrichment of damaging missense variants in genes related with axonal guidance signalling in sporadic Menierea€™s disease. Journal of Medical Genetics, 2020, 57, 82-88.	1.5	21
2444	A comparative analysis of genetic hearing loss phenotypes in European/American and Japanese populations. Human Genetics, 2020, 139, 1315-1323.	1.8	12
2445	Oligogenicity, C9orf72 expansion, and variant severity in ALS. Neurogenetics, 2020, 21, 227-242.	0.7	13
2446	Clinical utility of serial analysis of circulating tumour cells for detection of minimal residual disease of metastatic nasopharyngeal carcinoma. British Journal of Cancer, 2020, 123, 114-125.	2.9	14
2447	Defined lifestyle and germline factors predispose Asian populations to gastric cancer. Science Advances, 2020, 6, eaav9778.	4.7	31
2448	Clustering of known low and moderate risk alleles rather than a novel recessive higha€•risk gene in nona€• BRCA1 /2 sib trios affected with breast cancer. International Journal of Cancer, 2020, 147, 2708-2716.	2.3	2
2449	Constraint and conservation of paireda€•type homeodomains predicts the clinical outcome of missense variants of uncertain significance. Human Mutation, 2020, 41, 1407-1424.	1.1	2
2450	Knowledge-based analyses reveal new candidate genes associated with risk of hepatitis B virus related hepatocellular carcinoma. BMC Cancer, 2020, 20, 403.	1.1	6
2451	Identifying disease-causing mutations in genomes of single patients by computational approaches. Human Genetics, 2020, 139, 769-776.	1.8	12

#	Article	IF	Citations
2453	Challenges in the diagnosis and discovery of rare genetic disorders using contemporary sequencing technologies. Briefings in Functional Genomics, 2020, 19, 243-258.	1.3	27
2454	Whole Genome Analysis of Ovarian Granulosa Cell Tumors Reveals Tumor Heterogeneity and a High-Grade TP53-Specific Subgroup. Cancers, 2020, 12, 1308.	1.7	41
2455	Strategies for Functional Interrogation of Big Cancer Data Using Drosophila Cancer Models. International Journal of Molecular Sciences, 2020, 21, 3754.	1.8	5
2456	Variations in the TRPV1 gene are associated to exertional heat stroke. Journal of Science and Medicine in Sport, 2020, 23, 1021-1027.	0.6	7
2457	Expanding the clinical and genetic spectrum of ALPK3 variants: Phenotypes identified in pediatric cardiomyopathy patients and adults with heterozygous variants. American Heart Journal, 2020, 225, 108-119.	1.2	25
2458	Myeloproliferative Diseases as Possible Risk Factor for Development of Chronic Thromboembolic Pulmonary Hypertensionâ€"A Genetic Study. International Journal of Molecular Sciences, 2020, 21, 3339.	1.8	13
2459	Mutation severity spectrum of rare alleles in the human genome is predictive of disease type. PLoS Computational Biology, 2020, 16, e1007775.	1.5	11
2460	Genetic homozygosity in a diverse population: An experience of long QT syndrome. International Journal of Cardiology, 2020, 316, 117-124.	0.8	2
2461	Frequency of Usher gene mutations in non-syndromic hearing loss: higher variability of the Usher phenotype. Journal of Human Genetics, 2020, 65, 855-864.	1.1	6
2462	Population genetic models of GERP scores suggest pervasive turnover of constrained sites across mammalian evolution. PLoS Genetics, 2020, 16, e1008827.	1.5	65
2463	Pituispheres Contain Genetic Variants Characteristic to Pituitary Adenoma Tumor Tissue. Frontiers in Endocrinology, 2020, 11, 313.	1.5	5
2464	Novel compound heterozygous variants in NHLRC2 in a patient with FINCA syndrome. Journal of Human Genetics, 2020, 65, 911-915.	1.1	11
2465	Co-localization between Sequence Constraint and Epigenomic Information Improves Interpretation of Whole-Genome Sequencing Data. American Journal of Human Genetics, 2020, 106, 513-524.	2.6	3
2466	Landscape of multi-nucleotide variants in 125,748 human exomes and 15,708 genomes. Nature Communications, 2020, 11, 2539.	5.8	98
2467	A Systematic Literature Review of Whole Exome and Genome Sequencing Population Studies of Genetic Susceptibility to Cancer. Cancer Epidemiology Biomarkers and Prevention, 2020, 29, 1519-1534.	1.1	10
2468	Association of TIM-1 (T-Cell Immunoglobulin and Mucin Domain 1) With Incidence of Stroke. Arteriosclerosis, Thrombosis, and Vascular Biology, 2020, 40, 1777-1786.	1.1	5
2469	Computational Approaches for Unraveling the Effects of Variation in the Human Genome and Microbiome. Annual Review of Biomedical Data Science, 2020, 3, 411-432.	2.8	5
2470	The necdin interactome: evaluating the effects of amino acid substitutions and cell stress using proximity-dependent biotinylation (BioID) and mass spectrometry. Human Genetics, 2020, 139, 1513-1529.	1.8	4

#	Article	IF	Citations
2471	Childhood rare lung disease in the 21st century: â€œâ€•omicsâ€•technology advances accelerating discovery. Pediatric Pulmonology, 2020, 55, 1828-1837.	1.0	8
2472	Whole exome sequencing in patients with Williamsâ€“Beuren syndrome followed by disease modeling in mice points to four novel pathways that may modify stenosis risk. Human Molecular Genetics, 2020, 29, 2035-2050.	1.4	15
2473	Multi-omic analysis reveals significantly mutated genes and DDX3X as a sex-specific tumor suppressor in cutaneous melanoma. Nature Cancer, 2020, 1, 635-652.	5.7	26
2474	The role of rare compound heterozygous events in autism spectrum disorder. Translational Psychiatry, 2020, 10, 204.	2.4	2
2475	Mapping and characterization of structural variation in 17,795 human genomes. Nature, 2020, 583, 83-89.	13.7	194
2476	Whole-genome sequencing of patients with rare diseases in a national health system. Nature, 2020, 583, 96-102.	13.7	338
2477	Granularity of SERPINA1 alleles by DNA sequencing in CanCOLD. European Respiratory Journal, 2020, 56, 2000958.	3.1	13
2478	Long-read human genome sequencing and its applications. Nature Reviews Genetics, 2020, 21, 597-614.	7.7	542
2479	Hemizygous mutations in L1CAM in two unrelated male probands with childhood onset psychosis. Psychiatric Genetics, 2020, 30, 73-82.	0.6	2
2480	Origins and characterization of variants shared between databases of somatic and germline human mutations. BMC Bioinformatics, 2020, 21, 227.	1.2	14
2481	A Germline Mutation in the POT1 Gene Is a Candidate for Familial Non-Medullary Thyroid Cancer. Cancers, 2020, 12, 1441.	1.7	24
2482	SMAD6 variants in craniosynostosis: genotype and phenotype evaluation. Genetics in Medicine, 2020, 22, 1498-1506.	1.1	31
2483	Whole-Exome Sequencing-Based Approach for Germline Mutations in Patients with Inborn Errors of Immunity. Journal of Clinical Immunology, 2020, 40, 729-740.	2.0	20
2484	Biallelic variants in the RNA exosome gene EXOSC5 are associated with developmental delays, short stature, cerebellar hypoplasia and motor weakness. Human Molecular Genetics, 2020, 29, 2218-2239.	1.4	19
2485	Bartter syndrome representing digenic-based salt-losing tubulopathies presumably accelerated by renal insufficiency. CEN Case Reports, 2020, 9, 375-379.	0.5	0
2486	Coexpression enrichment analysis at the single-cell level reveals convergent defects in neural progenitor cells and their cell-type transitions in neurodevelopmental disorders. Genome Research, 2020, 30, 835-848.	2.4	25
2487	Genetic analysis of nucleotide-binding leucine-rich repeat (NLR) receptors in multiple sclerosis. Immunogenetics, 2020, 72, 381-385.	1.2	6
2488	Identifying genetic variants and pathways associated with extreme levels of fetal hemoglobin in sickle cell disease in Tanzania. BMC Medical Genetics, 2020, 21, 125.	2.1	9

#	Article	IF	Citations
2489	Genome-wide association study identifies genetic susceptibility loci and pathways of radiation-induced acute oral mucositis. Journal of Translational Medicine, 2020, 18, 224.	1.8	29
2490	Phen2Gene: rapid phenotype-driven gene prioritization for rare diseases. NAR Genomics and Bioinformatics, 2020, 2, lqaa032.	1.5	45
2491	Pitfalls in Single Clone CRISPR-Cas9 Mutagenesis to Fine-Map Regulatory Intervals. Genes, 2020, 11, 504.	1.0	6
2492	Nonâ€random distribution of deleterious mutations in the DNA and proteinâ€binding domains of IRF6 are associated with Van Der Woude syndrome. Molecular Genetics & Genomic Medicine, 2020, 8, e1355.	0.6	13
2493	The genomic and epigenomic evolutionary history of papillary renal cell carcinomas. Nature Communications, 2020, 11, 3096.	5.8	19
2494	A novel mutation in ABCD1 gene in a Filipino patient with adultâ€onset Xâ€linked ALD. Neurology and Clinical Neuroscience, 2020, 8, 329-331.	0.2	0
2495	Cumulative Influence of Inflammatory Response Genetic Variation on Long-Term Neurobehavioral Outcomes after Pediatric Traumatic Brain Injury Relative to Orthopedic Injury: An Exploratory Polygenic Risk Score. Journal of Neurotrauma, 2020, 37, 1491-1503.	1.7	10
2496	Identification of transmembrane protein 168 mutation in familial Brugada syndrome. FASEB Journal, 2020, 34, 6399-6417.	0.2	6
2497	A homozygous missense variant in CACNB4 encoding the auxiliary calcium channel beta4 subunit causes a severe neurodevelopmental disorder and impairs channel and non-channel functions. PLoS Genetics, 2020, 16, e1008625.	1.5	18
2498	Exploring the genetic pathogenicity of aortic dissection from 72 Han Chinese individuals using nextâ€generation sequencing. Clinical Genetics, 2020, 97, 704-711.	1.0	4
2499	An interpretable low-complexity machine learning framework for robust exome-based in-silico diagnosis of Crohnâ€™s disease patients. NAR Genomics and Bioinformatics, 2020, 2, lqaa011.	1.5	11
2500	Genomic Variants of Cytarabine Sensitivity Associated with Treatment-Related Mortality in Pediatric AML: A Report from the Children's Oncology Group. Clinical Cancer Research, 2020, 26, 2891-2897.	3.2	3
2501	The genetic architecture of the human cerebral cortex. Science, 2020, 367, .	6.0	450
2502	Inherited DNA Repair Gene Mutations in Men with Lethal Prostate Cancer. Genes, 2020, 11, 314.	1.0	16
2503	Emerging strategies to bridge the gap between pharmacogenomic research and its clinical implementation. Npj Genomic Medicine, 2020, 5, 9.	1.7	42
2504	8q24 genetic variation and comprehensive haplotypes altering familial risk of prostate cancer. Nature Communications, 2020, 11, 1523.	5.8	10
2505	Identification of Familial Hodgkin Lymphoma Predisposing Genes Using Whole Genome Sequencing. Frontiers in Bioengineering and Biotechnology, 2020, 8, 179.	2.0	12
2506	KCNN2 mutation in autosomalâ€dominant tremulous myoclonusâ€dystonia. European Journal of Neurology, 2020, 27, 1471-1477.	1.7	21

#	Article	IF	Citations
2507	Pathway Mutations in Breast Cancer Using Whole-Exome Sequencing. Oncology Research, 2020, 28, 107-116.	0.6	15
2508	<scp>Wolffâ€“Parkinsonâ€“White</scp> syndrome: De novo variants and evidence for mutational burden in genes associated with atrial fibrillation. American Journal of Medical Genetics, Part A, 2020, 182, 1387-1399.	0.7	14
2509	When transcripts matter: delineating between non-syndromic hearing loss DFNB32 and hearing impairment infertile male syndrome (HIIMS). Journal of Human Genetics, 2020, 65, 609-617.	1.1	2
2510	Identifying blood pressure loci whose effects are modulated by multiple lifestyle exposures. Genetic Epidemiology, 2020, 44, 629-641.	0.6	6
2511	Largeâ€scale in vitro functional testing and novel variant scoring via protein modeling provide insights into alkaline phosphatase activity in hypophosphatasia. Human Mutation, 2020, 41, 1250-1262.	1.1	32
2512	Improved Criteria for the Classification of Titin Variants in Inherited Skeletal Myopathies. Journal of Neuromuscular Diseases, 2020, 7, 153-166.	1.1	18
2513	Identification of a rare SEPT9 variant in a family with autosomal dominant Charcot-Marie-Tooth disease. BMC Medical Genetics, 2020, 21, 45.	2.1	5
2514	Novel and Known Gene-Smoking Interactions With cIMT Identified as Potential Drivers for Atherosclerosis Risk in West-African Populations of the AWI-Gen Study. Frontiers in Genetics, 2019, 10, 1354.	1.1	10
2515	Expanding Clinical Presentations Due to Variations in THOC2 mRNA Nuclear Export Factor. Frontiers in Molecular Neuroscience, 2020, 13, 12.	1.4	12
2516	Evolution of approaches to identify melanoma missing heritability. Expert Review of Molecular Diagnostics, 2020, 20, 523-531.	1.5	16
2517	Genomeâ€wide metaâ€analysis identified novel variant associated with hallux valgus in Caucasians. Journal of Foot and Ankle Research, 2020, 13, 11.	0.7	9
2518	The molecular landscape of osteogenesis imperfecta in a Brazilian tertiary service cohort. Osteoporosis International, 2020, 31, 1341-1352.	1.3	5
2519	Low Prevalence of GSC Gene Mutations in a Large Cohort of Predominantly Caucasian Patients with Hidradenitis Suppurativa. Journal of Investigative Dermatology, 2020, 140, 2085-2088.e14.	0.3	47
2520	Genome sequencing of human in vitro fertilisation embryos for pathogenic variation screening. Scientific Reports, 2020, 10, 3795.	1.6	15
2521	LIST-S2: taxonomy based sorting of deleterious missense mutations across species. Nucleic Acids Research, 2020, 48, W154-W161.	6.5	46
2522	KIAA1109 gene mutation in surviving patients with Alkuraya-KuÄinskas syndrome: a review of literature. BMC Medical Genetics, 2020, 21, 136.	2.1	10
2523	Genomic analyses implicate noncoding de novo variants in congenital heart disease. Nature Genetics, 2020, 52, 769-777.	9.4	97
2524	Evaluating heterogeneity in <scp>ASD</scp> symptomatology, cognitive ability, and adaptive functioning among 16p11.2 <scp>CNV</scp> carriers. Autism Research, 2020, 13, 1300-1310.	2.1	23

#	Article	IF	Citations
2525	CantÃº syndrome versus Zimmermann-Laband syndrome: Report of nine individuals with ABCC9 variants. European Journal of Medical Genetics, 2020, 63, 103996.	0.7	7
2526	Homozygote CRIM1 variant is associated with thiopurine-induced neutropenia in leukemic patients with both wildtype NUDT15 and TPMT. Journal of Translational Medicine, 2020, 18, 265.	1.8	3
2527	DLG5 variants are associated with multiple congenital anomalies including ciliopathy phenotypes. Journal of Medical Genetics, 2021, 58, 453-464.	1.5	10
2528	HEM1 deficiency disrupts mTORC2 and F-actin control in inherited immunodysregulatory disease. Science, 2020, 369, 202-207.	6.0	65
2529	Statistical methods for SNP heritability estimation and partition: A review. Computational and Structural Biotechnology Journal, 2020, 18, 1557-1568.	1.9	41
2530	The association between Single Nucleotide Polymorphisms of Klotho Gene and Mortality in Elderly Men: The MrOS Sweden Study. Scientific Reports, 2020, 10, 10243.	1.6	3
2531	Multi-trait Genome-Wide Analyses of the Brain Imaging Phenotypes in UK Biobank. Genetics, 2020, 215, 947-958.	1.2	10
2532	Genotypeâ€“Phenotype Correlations in a Spanish Cohort of 506 Families With Biallelic ABCA4 Pathogenic Variants. American Journal of Ophthalmology, 2020, 219, 195-204.	1.7	20
2533	Regulation of inflammation in diabetes: From genetics to epigenomics evidence. Molecular Metabolism, 2020, 41, 101041.	3.0	23
2534	A new paraplegin mutation in a patient with primary progressive multiple sclerosis. Multiple Sclerosis and Related Disorders, 2020, 44, 102302.	0.9	3
2535	Clinical interpretation of variants identified in RNU4ATAC, a non-coding spliceosomal gene. PLoS ONE, 2020, 15, e0235655.	1.1	8
2536	DeepWAS: Multivariate genotype-phenotype associations by directly integrating regulatory information using deep learning. PLoS Computational Biology, 2020, 16, e1007616.	1.5	54
2537	Identification and functional characterization of a novel heterozygous missense variant in the LPL associated with recurrent hypertriglyceridemiaâ€induced acute pancreatitis in pregnancy. Molecular Genetics & Genomic Medicine, 2020, 8, e1048.	0.6	11
2538	Modifier genes in SCN1Aâ€related epilepsy syndromes. Molecular Genetics & Genomic Medicine, 2020, 8, e1103.	0.6	16
2539	Germline variants in DNA repair genes associated with hereditary breast and ovarian cancer syndrome: analysis of a 21 gene panel in the Brazilian population. BMC Medical Genomics, 2020, 13, 21.	0.7	32
2540	Genetic associations between serum low LDL-cholesterol levels and variants in LDLR, APOB, PCSK9 andÂLDLRAP1 in African populations. PLoS ONE, 2020, 15, e0229098.	1.1	9
2541	Novel pathogenic alterations in pediatric and adult desmoid-type fibromatosis â€“ A systematic analysis of 204 cases. Scientific Reports, 2020, 10, 3368.	1.6	29
2542	Rhapsody: predicting the pathogenicity of human missense variants. Bioinformatics, 2020, 36, 3084-3092.	1.8	63

#	Article	IF	Citations
2543	Mechanosensitive Piezo1 ion channel protein (PIEZO1 gene): update and extended mutation analysis of hereditary xerocytosis in India. Annals of Hematology, 2020, 99, 715-727.	0.8	9
2544	De Novo Variants in SPOP Cause Two Clinically Distinct Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 405-411.	2.6	8
2545	Mechanistic insights into the deleterious roles of Nasu-Hakola disease associated TREM2 variants. Scientific Reports, 2020, 10, 3663.	1.6	24
2546	Nine newly identified individuals refine the phenotype associated with MYT1L mutations. American Journal of Medical Genetics, Part A, 2020, 182, 1021-1031.	0.7	19
2547	EEF1A2 mutations in epileptic encephalopathy/intellectual disability: Understanding the potential mechanism of phenotypic variation. Epilepsy and Behavior, 2020, 105, 106955.	0.9	11
2548	A polygenic predictor of treatment-resistant depression using whole exome sequencing and genome-wide genotyping. Translational Psychiatry, 2020, 10, 50.	2.4	33
2549	Exome Sequencing in Individuals with Isolated Biliary Atresia. Scientific Reports, 2020, 10, 2709.	1.6	17
2550	How We Got to CRISPR: The Dilemma of Being Human. Perspectives in Biology and Medicine, 2020, 63, 28-43.	0.3	11
2551	Neurological Involvement in Glycogen Storage Disease Type IXa due to PHKA2 Mutation. Canadian Journal of Neurological Sciences, 2020, 47, 400-403.	0.3	7
2552	Exome-Based Case-Control Analysis Highlights the Pathogenic Role of Ciliary Genes in Transposition of the Great Arteries. Circulation Research, 2020, 126, 811-821.	2.0	29
2553	Purging of highly deleterious mutations through severe bottlenecks in Alpine ibex. Nature Communications, 2020, 11, 1001.	5.8	147
2554	Massively parallel variant characterization identifies NUDT15 alleles associated with thiopurine toxicity. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 5394-5401.	3.3	95
2555	Deleterious variants in genes associated with bone mineral density are linked to susceptibility to periodontitis development. Meta Gene, 2020, 24, 100670.	0.3	2
2556	The genetics of situs inversus without primary ciliary dyskinesia. Scientific Reports, 2020, 10, 3677.	1.6	37
2557	Six yearsâ€™ experience with LipidSeq: clinical and research learnings from a hybrid, targeted sequencing panel for dyslipidemias. BMC Medical Genomics, 2020, 13, 23.	0.7	52
2558	Whole genome sequencing unveils genetic heterogeneity in optic nerve hypoplasia. PLoS ONE, 2020, 15, e0228622.	1.1	6
2559	Posterior column ataxia with retinitis pigmentosa (PCARP) in an Iranian patient associated with the FLVCR1 gene. Ophthalmic Genetics, 2020, 41, 90-92.	0.5	3
2560	A novel deep intronic SERPING1 variant as a cause of hereditary angioedema due to C1-inhibitor deficiency. Allergology International, 2020, 69, 443-449.	1.4	19

#	Article	IF	Citations
2561	Rewriting Human History and Empowering Indigenous Communities with Genome Editing Tools. Genes, 2020, 11, 88.	1.0	9
2562	Genetic Predisposition to High-Altitude Pulmonary Edema. High Altitude Medicine and Biology, 2020, 21, 28-36.	0.5	21
2563	Severe combined immunodeficiency caused by inositol-trisphosphate 3-kinase B (ITPKB) deficiency. Journal of Allergy and Clinical Immunology, 2020, 145, 1696-1699.e6.	1.5	6
2564	Germline cancer predisposition variants and pediatric glioma: a population-based study in California. Neuro-Oncology, 2020, 22, 864-874.	0.6	24
2565	De novo variants in exomes of congenital heart disease patients identify risk genes and pathways. Genome Medicine, 2020, 12, 9.	3.6	43
2566	The polygenic nature of mild-to-moderate hypertriglyceridemia. Journal of Clinical Lipidology, 2020, 14, 28-34.e2.	0.6	32
2567	Phenotypic and genotypic characterization of families with complex intellectual disability identified pathogenic genetic variations in known and novel disease genes. Scientific Reports, 2020, 10, 968.	1.6	8
2568	In-silico Analysis of NF1 Missense Variants in ClinVar: Translating Variant Predictions into Variant Interpretation and Classification. International Journal of Molecular Sciences, 2020, 21, 721.	1.8	9
2569	Genome-wide rare variant analysis for thousands of phenotypes in over 70,000 exomes from two cohorts. Nature Communications, 2020, 11, 542.	5.8	101
2570	Brown-Vialetto-Van Laere syndrome and Fazio-Londe syndrome: A novel mutation and in silico analyses. Journal of Clinical Neuroscience, 2020, 72, 342-349.	0.8	5
2571	A proactive genotype-to-patient-phenotype map for cystathionine beta-synthase. Genome Medicine, 2020, 12, 13.	3.6	45
2572	Combined burden and functional impact tests for cancer driver discovery using DriverPower. Nature Communications, 2020, 11, 734.	5.8	39
2573	Genotype-phenotype correlation and description of two novel mutations in Iranian patients with glycogen storage disease 1b (GSD1b). Orphanet Journal of Rare Diseases, 2020, 15, 35.	1.2	4
2574	Genetic Analyses in Dent Disease and Characterization of CLCN5 Mutations in Kidney Biopsies. International Journal of Molecular Sciences, 2020, 21, 516.	1.8	17
2575	Statistical Methods in Genome-Wide Association Studies. Annual Review of Biomedical Data Science, 2020, 3, 265-288.	2.8	6
2576	Variants in CAPZA2, a member of an F-actin capping complex, cause intellectual disability and developmental delay. Human Molecular Genetics, 2020, 29, 1537-1546.	1.4	15
2577	De novo variants in CUL3 are associated with global developmental delays with or without infantile spasms. Journal of Human Genetics, 2020, 65, 727-734.	1.1	23
2578	Contribution of common and rare damaging variants in familial forms of bipolar disorder and phenotypic outcome. Translational Psychiatry, 2020, 10, 124.	2.4	5

#	Article	IF	Citations
2579	DLG2 variants in patients with pubertal disorders. Genetics in Medicine, 2020, 22, 1329-1337.	1.1	7
2580	Common genetic variation in obesity, lipid transfer genes and risk of Metabolic Syndrome: Results from IDEFICS/I.Family study and meta-analysis. Scientific Reports, 2020, 10, 7189.	1.6	23
2581	Using an integrative machine learning approach utilising homology modelling to clinically interpret genetic variants: CACNA1F as an exemplar. European Journal of Human Genetics, 2020, 28, 1274-1282.	1.4	11
2582	Multi-model functionalization of disease-associated PTEN missense mutations identifies multiple molecular mechanisms underlying protein dysfunction. Nature Communications, 2020, 11, 2073.	5.8	34
2583	CAPN1 and hereditary spastic paraplegia: a novel variant in an Iranian family and overview of the genotype-phenotype correlation. International Journal of Neuroscience, 2020, 131, 1-13.	0.8	7
2584	Structural analysis of pathogenic missense mutations in GABRA2 and identification of a novel de novo variant in the desensitization gate. Molecular Genetics & Genomic Medicine, 2020, 8, e1106.	0.6	9
2585	MonoallelicÂloss-of-function THPOÂvariants cause heritable thrombocytopenia. Blood Advances, 2020, 4, 920-924.	2.5	10
2586	Understanding oncogenicity of cancer driver genes and mutations in the cancer genomics era. FEBS Letters, 2020, 594, 4233-4246.	1.3	20
2587	CACNA1H variants are not a cause of monogenic epilepsy. Human Mutation, 2020, 41, 1138-1144.	1.1	19
2588	Missing heritability in Parkinsonâ€™s disease: the emerging role of non-coding genetic variation. Journal of Neural Transmission, 2020, 127, 729-748.	1.4	27
2589	The contribution of rare genetic variants to the pathogenesis of polycystic ovary syndrome. Current Opinion in Endocrine and Metabolic Research, 2020, 12, 26-32.	0.6	21
2590	Machine learning, the kidney, and genotypeâ€“phenotype analysis. Kidney International, 2020, 97, 1141-1149.	2.6	23
2591	Piezo1 is required for outflow tract and aortic valve development.. Journal of Molecular and Cellular Cardiology, 2020, 143, 51-62.	0.9	44
2592	Whole exome sequencing in ADHD trios from single and multi-incident families implicates new candidate genes and highlights polygenic transmission. European Journal of Human Genetics, 2020, 28, 1098-1110.	1.4	13
2593	A novel mutation in TARDBP segregates with amyotrophic lateral sclerosis in a large family with early onset and fast progression. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2020, 21, 280-285.	1.1	0
2594	Integrated Analysis of Summary Statistics to Identify Pleiotropic Genes and Pathways for the Comorbidity of Schizophrenia and Cardiometabolic Disease. Frontiers in Psychiatry, 2020, 11, 256.	1.3	24
2595	An Improved Phenotype-Driven Tool for Rare Mendelian Variant Prioritization: Benchmarking Exomiser on Real Patient Whole-Exome Data. Genes, 2020, 11, 460.	1.0	42
2596	Multi-level evidence of an allelic hierarchy of USH2A variants in hearing, auditory processing and speech/language outcomes. Communications Biology, 2020, 3, 180.	2.0	6

#	Article	IF	Citations
2597	Compound Heterozygous Variants in Pediatric Cancers: A Systematic Review. Frontiers in Genetics, 2020, 11, 493.	1.1	13
2598	Excess of singleton loss-of-function variants in Parkinsonâ€™s disease contributes to genetic risk. Journal of Medical Genetics, 2020, 57, 617-623.	1.5	10
2599	A homozygous UBA5 pathogenic variant causes a fatal congenital neuropathy. Journal of Medical Genetics, 2020, 57, 835-842.	1.5	16
2600	TLmutation: Predicting the Effects of Mutations Using Transfer Learning. Journal of Physical Chemistry B, 2020, 124, 3845-3854.	1.2	21
2601	Predicting target genes of non-coding regulatory variants with IRT. Bioinformatics, 2020, 36, 4440-4448.	1.8	6
2602	Clinicoâ€Genetic, Imaging and Molecular Delineation of <scp>COQ8A</scp>â€Ataxia: A Multicenter Study of 59 Patients. Annals of Neurology, 2020, 88, 251-263.	2.8	52
2603	Non-coding and Loss-of-Function Coding Variants in TET2 are Associated with Multiple Neurodegenerative Diseases. American Journal of Human Genetics, 2020, 106, 632-645.	2.6	50
2604	A catalog of the pathogenic mutations of LDL receptor gene in Japanese familial hypercholesterolemia. Journal of Clinical Lipidology, 2020, 14, 346-351.e9.	0.6	20
2605	CScape-somatic: distinguishing driver and passenger point mutations in the cancer genome. Bioinformatics, 2020, 36, 3637-3644.	1.8	19
2606	Loss of ap4s1 in zebrafish leads to neurodevelopmental defects resembling spastic paraplegia 52. Annals of Clinical and Translational Neurology, 2020, 7, 584-589.	1.7	15
2607	dbMTS: A comprehensive database of putative human microRNA target site SNVs and their functional predictions. Human Mutation, 2020, 41, 1123-1130.	1.1	11
2608	Collagen VI-related limb-girdle syndrome caused by frequent mutation in COL6A3 gene with conflicting reports of pathogenicity. Neuromuscular Disorders, 2020, 30, 483-491.	0.3	3
2609	Infantile hypertrophic pyloric stenosis in patients with esophageal atresia. Birth Defects Research, 2020, 112, 670-687.	0.8	1
2610	Biallelic Mutations in the LSR Gene Cause a Novel Type of Infantile Intrahepatic Cholestasis. Journal of Pediatrics, 2020, 221, 251-254.	0.9	11
2611	Prot2HG: a database of protein domains mapped to the human genome. Database: the Journal of Biological Databases and Curation, 2020, 2020, .	1.4	17
2612	Reporting a rare form of myopathy, myopathy with extrapyramidal signs, in an Iranian family using next generation sequencing: a case report. BMC Medical Genetics, 2020, 21, 77.	2.1	11
2613	Foetal onset of EIF2B related disorder in two siblings: cerebellar hypoplasia with absent Bergmann glia and severe hypomyelination. Acta Neuropathologica Communications, 2020, 8, 48.	2.4	4
2614	Heterozygous STUB1 missense variants cause ataxia, cognitive decline, and STUB1 mislocalization. Neurology: Genetics, 2020, 6, 1-13.	0.9	19

#	Article	IF	Citations
2615	Relaxed constraint and functional divergence of the progesterone receptor (PGR) in the human stem-lineage. PLoS Genetics, 2020, 16, e1008666.	1.5	9
2616	Genetic landscape of preterm birth due to cervical insufficiency: Comprehensive gene analysis and patient next-generation sequencing data interpretation. PLoS ONE, 2020, 15, e0230771.	1.1	14
2617	Whole Exome Sequencing of Highly Aggregated Lung Cancer Families Reveals Linked Loci for Increased Cancer Risk on Chromosomes 12q, 7p, and 4q. Cancer Epidemiology Biomarkers and Prevention, 2020, 29, 434-442.	1.1	11
2618	The Occurrence of FUS Mutations in Pediatric Amyotrophic Lateral Sclerosis: A Case Report and Review of the Literature. Journal of Child Neurology, 2020, 35, 556-562.	0.7	23
2619	Current genetic landscape in common variable immune deficiency. Blood, 2020, 135, 656-667.	0.6	109
2620	The mutational burden and oligogenic inheritance in Klippel-Feil syndrome. BMC Musculoskeletal Disorders, 2020, 21, 220.	0.8	15
2621	Mutations in COL1A1/A2 and CREB3L1 are associated with oligodontia in osteogenesis imperfecta. Orphanet Journal of Rare Diseases, 2020, 15, 80.	1.2	10
2622	Characterisation of genetic regulatory effects for osteoporosis risk variants in human osteoclasts. Genome Biology, 2020, 21, 80.	3.8	36
2623	Loss-of-function tolerance of enhancers in the human genome. PLoS Genetics, 2020, 16, e1008663.	1.5	12
2624	Phenogenon: Gene to phenotype associations for rare genetic diseases. PLoS ONE, 2020, 15, e0230587.	1.1	6
2625	Is Gene-Size an Issue for the Diagnosis of Skeletal Muscle Disorders?. Journal of Neuromuscular Diseases, 2020, 7, 203-216.	1.1	9
2626	Exome-Wide Association Study Reveals Several Susceptibility Genes and Pathways Associated With Acute Coronary Syndromes in Han Chinese. Frontiers in Genetics, 2020, 11, 336.	1.1	3
2627	Resources for functional genomic studies of health and development in nonhuman primates. American Journal of Physical Anthropology, 2020, 171, 174-194.	2.1	7
2628	Genetic diagnosis of autoinflammatory disease patients using clinical exome sequencing. European Journal of Medical Genetics, 2020, 63, 103920.	0.7	15
2629	Thrombocytosis in an infant with a TRPV4 mutation: a case report. Platelets, 2021, 32, 429-431.	1.1	3
2630	Identification of a homozygous recessive variant in <i>PTGS1</i> resulting in a congenital aspirin-like defect in platelet function. Haematologica, 2021, 106, 1423-1432.	1.7	7
2631	Whole Exome Sequencing reveals NOTCH1 mutations in anaplastic large cell lymphoma and points to Notch both as a key pathway and a potential therapeutic target. Haematologica, 2021, 106, 1693-1704.	1.7	40
2632	Prevalence and associated phenotypes of DUSP6, IL17RD and SPRY4 variants in a large Chinese cohort with isolated hypogonadotropic hypogonadism. Journal of Medical Genetics, 2021, 58, 66-72.	1.5	5

#	Article	IF	Citations
2633	Emerging Methods and Resources for BiologicalÂInterrogation of Neuropsychiatric Polygenic Signal. Biological Psychiatry, 2021, 89, 41-53.	0.7	38
2634	Genome-wide Association Analysis of Parkinsonâ€™s Disease and Schizophrenia Reveals Shared Genetic Architecture and Identifies Novel Risk Loci. Biological Psychiatry, 2021, 89, 227-235.	0.7	53
2635	PredCID: prediction of driver frameshift indels in human cancer. Briefings in Bioinformatics, 2021, 22, .	3.2	29
2636	Variant Calling in Next Generation Sequencing Data. , 2021, , 129-140.		0
2637	PIGH deficiency can be associated with severe neurodevelopmental and skeletal manifestations. Clinical Genetics, 2021, 99, 313-317.	1.0	7
2638	A RIPOR2 in-frame deletion is a frequent and highly penetrant cause of adult-onset hearing loss. Journal of Medical Genetics, 2021, 58, 96-104.	1.5	14
2639	Novel candidate genes in esophageal atresia/tracheoesophageal fistula identified by exome sequencing. European Journal of Human Genetics, 2021, 29, 122-130.	1.4	17
2640	Genetic and functional analysis of KIF5A variants in Japanese patients with sporadic amyotrophic lateral sclerosis. Neurobiology of Aging, 2021, 97, 147.e11-147.e17.	1.5	11
2641	Clinically and biologically relevant subgroups of Wilms tumour defined by genomic and epigenomic analyses. British Journal of Cancer, 2021, 124, 437-446.	2.9	16
2642	Identifying rare, medically relevant variation via population-based genomic screening in Alabama: opportunities and pitfalls. Genetics in Medicine, 2021, 23, 280-288.	1.1	9
2643	Prospective phenotyping of long-term survivors of generalized arterial calcification of infancy (GACI). Genetics in Medicine, 2021, 23, 396-407.	1.1	44
2644	Frequency and molecular characteristics of PALB2 â€associated cancers in Russian patients. International Journal of Cancer, 2021, 148, 203-210.	2.3	9
2645	Combined immunodeficiency caused by a novel homozygous NFKB1 mutation. Journal of Allergy and Clinical Immunology, 2021, 147, 727-733.e2.	1.5	13
2646	CSVS, a crowdsourcing database of the Spanish population genetic variability. Nucleic Acids Research, 2021, 49, D1130-D1137.	6.5	34
2647	Contribution of homozygous and compound heterozygous missense mutations in VWA2 to Alzheimerâ€™s disease. Neurobiology of Aging, 2021, 99, 100.e17-100.e23.	1.5	5
2648	Germline testing for homologous recombination repair genesâ€”opportunities and challenges. Genes Chromosomes and Cancer, 2021, 60, 332-343.	1.5	7
2649	BAX 335 hemophilia B gene therapy clinical trial results: potential impact of CpG sequences on gene expression. Blood, 2021, 137, 763-774.	0.6	94
2650	Multi-ethnic GWAS and meta-analysis of sleep quality identify MPP6 as a novel gene that functions in sleep center neurons. Sleep, 2021, 44, .	0.6	5

#	Article	IF	Citations
2651	Integration of multiomic annotation data to prioritize and characterize inflammation and immuneâ€related risk variants in squamous cell lung cancer. Genetic Epidemiology, 2021, 45, 99-114.	0.6	7
2652	Disease-specific variant pathogenicity prediction significantly improves variant interpretation in inherited cardiac conditions. Genetics in Medicine, 2021, 23, 69-79.	1.1	39
2653	Genotypeâ€“Phenotype Associations in 72 Adults with Suspected ALPL-Associated Hypophosphatasia. Calcified Tissue International, 2021, 108, 288-301.	1.5	16
2654	Detection and validation of novel mutations in MERTK in a simplex case of retinal degeneration using WGS and hiPSCâ€“RPEs model. Human Mutation, 2021, 42, 189-199.	1.1	5
2655	Predictive significance of selected gene mutations in relapsed and refractory chronic lymphocytic leukemia patients treated with ibrutinib. European Journal of Haematology, 2021, 106, 320-326.	1.1	2
2656	Familial periodic paralysis associated with a rare KCNJ5 variant that supposed to have incomplete penetrance. Brain and Development, 2021, 43, 470-474.	0.6	3
2657	INSAID Variant Classification and Eurofever Criteria Guide Optimal Treatment Strategy in Patients with TRAPS: Data from the Eurofever Registry. Journal of Allergy and Clinical Immunology: in Practice, 2021, 9, 783-791.e4.	2.0	16
2658	Mutation analysis of MFSD8 in an amyotrophic lateral sclerosis cohort from mainland China. European Journal of Neuroscience, 2021, 53, 1197-1206.	1.2	2
2659	Genetic markers for depressive disorders with earlier age at onset. Progress in Neuro-Psychopharmacology and Biological Psychiatry, 2021, 108, 110176.	2.5	4
2660	OncoVar: an integrated database and analysis platform for oncogenic driver variants in cancers. Nucleic Acids Research, 2021, 49, D1289-D1301.	6.5	64
2661	Variation analysis of anti-MÃ¼llerian hormone gene in Chinese women with polycystic ovary syndrome. Endocrine, 2021, 72, 287-293.	1.1	4
2662	Severe course with lethal hepatocellular injury and skeletal muscular dysgenesis in a neonate with infantile liver failure syndrome type 1 caused by novel <scp>LARS1</scp> mutations. American Journal of Medical Genetics, Part A, 2021, 185, 866-870.	0.7	4
2663	Identification of a novel de novo KMT2B variant in a Greek dystonia patient via exome sequencing genotypeâ€“phenotype correlations of all published cases. Molecular Biology Reports, 2021, 48, 371-379.	1.0	3
2664	Computational Tools for Causal Inference in Genetics. Cold Spring Harbor Perspectives in Medicine, 2021, 11, a039248.	2.9	3
2665	Genetic Predictors of Severe Skin Toxicity in Patients with Stage III Colon Cancer Treated with Cetuximab: NCCTG N0147 (Alliance). Cancer Epidemiology Biomarkers and Prevention, 2021, 30, 404-411.	1.1	1
2666	Epidemiological and ES cellâ€based functional evaluation of BRCA2 variants identified in families with breast cancer. Human Mutation, 2021, 42, 200-212.	1.1	4
2667	Germline variants of DNA repair genes in early onset mantle cell lymphoma. Oncogene, 2021, 40, 551-563.	2.6	9
2668	The burden of rare damaging variants in hereditary atypical parkinsonism genes is increased in patients with Parkinsonâ€™s disease. Neurobiology of Aging, 2021, 100, 118.e5-118.e13.	1.5	2

#	Article	IF	Citations
2669	New variants and in silico analyses in GRK1 associated Oguchi disease. Human Mutation, 2021, 42, 164-176.	1.1	7
2670	Functional and clinical implications of genetic structure in 1686 Italian exomes. Human Mutation, 2021, 42, 272-289.	1.1	5
2671	Biallelic ZNF335 mutations cause basal ganglia abnormality with progressive cerebral/cerebellar atrophy. Journal of Neurogenetics, 2021, 35, 23-28.	0.6	2
2672	JEM: A joint test to estimate the effect of multiple genetic variants on DNA methylation. Genetic Epidemiology, 2021, 45, 280-292.	0.6	0
2673	Shared Genetics of Multiple System Atrophy and Inflammatory Bowel Disease. Movement Disorders, 2021, 36, 449-459.	2.2	16
2674	Whole exome sequencing reveals pathogenic variants in MYO3A, MYO15A and COL9A3 and differential frequencies in ancestral alleles in hearing impairment genes among individuals from Cameroon. Human Molecular Genetics, 2021, 29, 3729-3743.	1.4	9
2675	Missense mutations in CASK, coding for the calciumâ€/calmodulinâ€dependent serine protein kinase, interfere with neurexin binding and neurexinâ€induced oligomerization. Journal of Neurochemistry, 2021, 157, 1331-1350.	2.1	14
2676	De novo variants in CELF2 that disrupt the nuclear localization signal cause developmental and epileptic encephalopathy. Human Mutation, 2021, 42, 66-76.	1.1	16
2677	The qBED track: a novel genome browser visualization for point processes. Bioinformatics, 2021, 37, 1168-1170.	1.8	4
2678	Increased Burden of Rare Sequence Variants in GnRH-Associated Genes in Women With Hypothalamic Amenorrhea. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e1441-e1452.	1.8	13
2679	Genomic analysis of lowâ€grade serous ovarian carcinoma to identify key drivers and therapeutic vulnerabilities. Journal of Pathology, 2021, 253, 41-54.	2.1	54
2680	Comprehensive functional annotation of susceptibility variants identifies genetic heterogeneity between lung adenocarcinoma and squamous cell carcinoma. Frontiers of Medicine, 2021, 15, 275-291.	1.5	21
2681	Aberrant expression of PAX6 gene associated with classical aniridia: identification and functional characterization of novel noncoding mutations. Journal of Human Genetics, 2021, 66, 333-338.	1.1	9
2682	Exome sequencing of 112 trios identifies recessive genetic variants in brain arteriovenous malformations. Journal of NeuroInterventional Surgery, 2021, 13, 568-573.	2.0	14
2683	Molecular dynamics simulations for genetic interpretation in protein coding regions: where we are, where to go and when. Briefings in Bioinformatics, 2021, 22, 3-19.	3.2	30
2684	Precision Medicine and Informatics. , 2021, , 941-966.		0
2685	Comprehensive Mutation Analysis and Report of 12 Novel Mutations in a Cohort of Patients with Spinal Muscular Atrophy in Iran. Journal of Molecular Neuroscience, 2021, 71, 2281-2298.	1.1	9
2686	Clustering suicidal phenotypes and genetic associations with brain-derived neurotrophic factor in patients with substance use disorders. Translational Psychiatry, 2021, 11, 72.	2.4	4

#	Article	IF	Citations
2687	Detecting Causal Variants in Mendelian Disorders Using Whole-Genome Sequencing. Methods in Molecular Biology, 2021, 2243, 1-25.	0.4	3
2688	Investigating the causal role of MRE11A p.E506* in breast and ovarian cancer. Scientific Reports, 2021, 11, 2409.	1.6	5
2689	Genomic diversity and post-admixture adaptation in the Uyghurs. National Science Review, 2022, 9, nwab124.	4.6	20
2690	A rare coding mutation in the MAST2 gene causes venous thrombosis in a French family with unexplained thrombophilia: The Breizh MAST2 Arg89Gln variant. PLoS Genetics, 2021, 17, e1009284.	1.5	2
2691	GPCards: An integrated database of genotypeâ€“phenotype correlations in human genetic diseases. Computational and Structural Biotechnology Journal, 2021, 19, 1603-1611.	1.9	5
2692	Isobutyrylâ€CoA dehydrogenase deficiency associated with autism in a girl without an alternative genetic diagnosis by trio whole exome sequencing: A case report. Molecular Genetics & Genomic Medicine, 2021, 9, e1595.	0.6	5
2693	<i>FLNC</i> and <i>MYLK2</i> Gene Mutations in a Chinese Family with Different Phenotypes of Cardiomyopathy. International Heart Journal, 2021, 62, 127-134.	0.5	6
2694	Prevalence of MYOC risk variants for glaucoma in different populations. Acta Ophthalmologica, 2021, 99, e1090-e1097.	0.6	7
2695	Network propagation of rare variants in Alzheimerâ€™s disease reveals tissue-specific hub genes and communities. PLoS Computational Biology, 2021, 17, e1008517.	1.5	1
2696	Sharing genetic variants with the NGS pipeline is essential for effective genomic data sharing and reproducibility in health information exchange. Scientific Reports, 2021, 11, 2268.	1.6	1
2697	Novel Compound Heterozygous Mutations in Two Families With Bernardâ€“Soulier Syndrome. Frontiers in Pediatrics, 2020, 8, 589812.	0.9	4
2698	Large mosaic copy number variations confer autism risk. Nature Neuroscience, 2021, 24, 197-203.	7.1	36
2699	A semisupervised model to predict regulatory effects of genetic variants at single nucleotide resolution using massively parallel reporter assays. Bioinformatics, 2021, , .	1.8	2
2700	Novel candidates of pathogenic variants of the BRCA1 and BRCA2 genes from a dataset of 3,552 Japanese whole genomes (3.5KJPNv2). PLoS ONE, 2021, 16, e0236907.	1.1	7
2702	Specific Cognitive Changes due to Hippocalcin Alterations? A Novel Familial Homozygous Hippocalcin Variant Associated with Inherited Dystonia and Altered Cognition. Neuropediatrics, 2021, 52, 377-382.	0.3	6
2703	Functional evidence (II) protein and enzyme function. , 2021, , 145-168.		0
2704	BCGitis as the primary manifestation of chronic granulomatous disease. IDCases, 2021, 23, e01038.	0.4	5
2707	Hunting for the perfect test: Neuromuscular diagnosis in the age of genomic bounty. Muscle and Nerve, 2021, 63, 282-284.	1.0	1

#	Article	IF	Citations
2709	Putative second hit rare genetic variants in families with seemingly GBA-associated Parkinsonâ€™s disease. Npj Genomic Medicine, 2021, 6, 2.	1.7	11
2710	Disease category-specific annotation of variants using an ensemble learning framework. Briefings in Bioinformatics, 2022, 23, .	3.2	7
2711	Prioritization of regulatory variants with tissue-specific function in the non-coding regions of human genome. Nucleic Acids Research, 2022, 50, e6-e6.	6.5	13
2712	Independent and cumulative coeliac disease-susceptibility loci are associated with distinct disease phenotypes. Journal of Human Genetics, 2021, 66, 613-623.	1.1	11
2713	MVP predicts theÂpathogenicity of missense variants by deep learning. Nature Communications, 2021, 12, 510.	5.8	85
2714	Genetic loci shared between major depression and intelligence with mixed directions of effect. Nature Human Behaviour, 2021, 5, 795-801.	6.2	23
2715	Biallelic SORD pathogenic variants cause Chinese patients with distal hereditary motor neuropathy. Npj Genomic Medicine, 2021, 6, 1.	1.7	32
2716	Alignment-free method for functional annotation of amino acid substitutions: Application on epigenetic factors involved in hematologic malignancies. PLoS ONE, 2021, 16, e0244948.	1.1	0
2717	Probing the aggregated effects of purifying selection per individual on 1,380 medical phenotypes in the UK Biobank. PLoS Genetics, 2021, 17, e1009337.	1.5	2
2718	Rare missense variant inMSH4associated with primary gonadal failure in both 46, XX and 46, XY individuals. Human Reproduction, 2021, 36, 1134-1145.	0.4	18
2719	A loss-of-function variant in DNA mismatch repair gene MLH3 underlies severe oligozoospermia. Journal of Human Genetics, 2021, 66, 725-730.	1.1	11
2720	Functional Network Profiles in ARSACS Disclosed by Aptamer-Based Proteomic Technology. Frontiers in Neurology, 2020, 11, 603774.	1.1	9
2721	Massively parallel functional testing of MSH2 missense variants conferring Lynch syndrome risk. American Journal of Human Genetics, 2021, 108, 163-175.	2.6	66
2722	Whole exome sequencing and system biology analysis support the "two-hit" mechanism in the onset of Ameloblastoma. Medicina Oral, Patologia Oral Y Cirugia Bucal, 2021, 26, e510-e517.	0.7	4
2723	Genetics of Cellular Immunodeficiencies. Rare Diseases of the Immune System, 2021, , 5-24.	0.1	0
2724	Exploring the Genetic Association of the ABAT Gene with Alzheimerâ€™s Disease. Molecular Neurobiology, 2021, 58, 1894-1903.	1.9	7
2725	Molecular Genetics in the Next Generation Sequencing Era. , 2021, , 215-230.		0
2726	Genetics of agenesis/hypoplasia of the uterus and vagina: narrowing down the number of candidate genes for Mayerâ€“Rokitanskyâ€“KÃ¼sterâ€“Hauser Syndrome. Human Genetics, 2021, 140, 667-680.	1.8	16

#	Article	IF	Citations
2727	Hearing loss. , 2021, , 305-322.		2
2728	R869C mutation in molecular motor KIF17 gene is involved in dementia with Lewy bodies. Alzheimer's and Dementia: Diagnosis, Assessment and Disease Monitoring, 2021, 13, e12143.	1.2	1
2729	Understanding protein structural changes for oncogenic missense variants. Heliyon, 2021, 7, e06013.	1.4	5
2730	Integrating Evolutionary Genetics to Medical Genomics: Evolutionary Approaches to Investigate Disease-Causing Variants. , 0, , .		0
2731	Whole Exome Sequencing Identifies APCDD1 and HDAC5 Genes as Potentially Cancer Predisposing in Familial Colorectal Cancer. International Journal of Molecular Sciences, 2021, 22, 1837.	1.8	6
2733	Genome-wide meta-analysis identifies 127 open-angle glaucoma loci with consistent effect across ancestries. Nature Communications, 2021, 12, 1258.	5.8	196
2734	Prognostic value of integrated cytogenetic, somatic variation, and copy number variation analyses in Korean patients with newly diagnosed multiple myeloma. PLoS ONE, 2021, 16, e0246322.	1.1	5
2735	Genome-wide association study identifies novel susceptibility loci for KIT D816V positive mastocytosis. American Journal of Human Genetics, 2021, 108, 284-294.	2.6	12
2736	First evidence of involvement of TBC1D25 in causing human male infertility. European Journal of Medical Genetics, 2021, 64, 104142.	0.7	8
2737	Clinical characterization and identification of rare genetic variants in atypical hemolytic uremic syndrome: A Swedish retrospective observational study. Therapeutic Apheresis and Dialysis, 2021, 25, 988-1000.	0.4	4
2738	Forkâ€shaped mandibular incisors as a novel phenotype of LRP5 â€associated disorder. American Journal of Medical Genetics, Part A, 2021, 185, 1544-1549.	0.7	1
2739	A founder mutation in the <scp>PLPBP</scp> gene in families from <scp>Saguenayâ€Lacâ€Stâ€Jean</scp> region affected by a pyridoxineâ€dependent epilepsy. JIMD Reports, 2021, 59, 32-41.	0.9	7
2740	CADD-Spliceâ€”improving genome-wide variant effect prediction using deep learning-derived splice scores. Genome Medicine, 2021, 13, 31.	3.6	375
2741	Replication of HLA class II locus association with susceptibility to podoconiosis in three Ethiopian ethnic groups. Scientific Reports, 2021, 11, 3285.	1.6	5
2743	Whole genome sequencing of 45 Japanese patients with intellectual disability. American Journal of Medical Genetics, Part A, 2021, 185, 1468-1480.	0.7	13
2744	Perturbations of genes essential for MÃ¼llerian duct and WÃ¶lffian duct development in Mayer-Rokitansky-KÃ¼ster-Hauser syndrome. American Journal of Human Genetics, 2021, 108, 337-345.	2.6	41
2745	The population genomics of adaptive loss of function. Heredity, 2021, 126, 383-395.	1.2	33
2746	A multilayered post-GWAS assessment on genetic susceptibility to pancreatic cancer. Genome Medicine, 2021, 13, 15.	3.6	15

#	Article	IF	Citations
2747	Common Treatment, Common Variant: Evolutionary Prediction of Functional Pharmacogenomic Variants. Journal of Personalized Medicine, 2021, 11, 131.	1.1	8
2748	PopDel identifies medium-size deletions simultaneously in tens of thousands of genomes. Nature Communications, 2021, 12, 730.	5.8	9
2749	A non-invasive method to generate induced pluripotent stem cells from primate urine. Scientific Reports, 2021, 11, 3516.	1.6	24
2751	DeepCAPE: A Deep Convolutional Neural Network for the Accurate Prediction of Enhancers. Genomics, Proteomics and Bioinformatics, 2021, 19, 565-577.	3.0	17
2752	Hirschsprung Disease in an Infant with L1 syndrome: Report of a New Case and a novel L1CAM variant. Clinical Case Reports (discontinued), 2021, 9, 1518-1523.	0.2	4
2753	Contribution of rare homozygous and compound heterozygous VPS13C missense mutations to dementia with Lewy bodies and Parkinsonâ€™s disease. Acta Neuropathologica Communications, 2021, 9, 25.	2.4	23
2755	eTumorMetastasis: A Network-based Algorithm Predicts Clinical Outcomes Using Whole-exome Sequencing Data of Cancer Patients. Genomics, Proteomics and Bioinformatics, 2021, 19, 973-985.	3.0	6
2756	Characterization of Hepatitis B Virus Integrations Identified in Hepatocellular Carcinoma Genomes. Viruses, 2021, 13, 245.	1.5	6
2758	Association of De Novo RNF213 Variants With Childhood Onset Moyamoya Disease and Diffuse Occlusive Vasculopathy. Neurology, 2021, 96, e1783-e1791.	1.5	21
2760	Customised targeted massively parallel sequencing enables more precise diagnosis of patients with epilepsy. Internal Medicine Journal, 2022, 52, 1174-1184.	0.5	4
2761	Mutational landscape and potential therapeutic targets for sporadic pancreatic neuroendocrine tumors based on target nextâ€generation sequencing. Experimental and Therapeutic Medicine, 2021, 21, 415.	0.8	1
2762	Identification of novel, clonally stable, somatic mutations targeting transcription factors PAX5 and NKX2-3, the epigenetic regulator LRIF1, and BRAF in a case of atypical B-cell chronic lymphocytic leukemia harboring a t(14;18)(q32;q21). Journal of Physical Education and Sports Management, 2021, 7, a005934.	0.5	1
2763	Next Generation Exome Sequencing of Pediatric Asthma Identifies Rare and Novel Variants in Candidate Genes. Disease Markers, 2021, 2021, 1-10.	0.6	6
2764	A case of retinitis pigmentosa homozygous for a rare CNGA1 causal variant. Scientific Reports, 2021, 11, 4681.	1.6	6
2765	From chemoproteomicâ€detected amino acids to genomic coordinates: insights into precise multiâ€omic data integration. Molecular Systems Biology, 2021, 17, e9840.	3.2	17
2766	Auto-antibodies to type I IFNs can underlie adverse reactions to yellow fever live attenuated vaccine. Journal of Experimental Medicine, 2021, 218, .	4.2	130
2767	Biallelic variants in COPB1 cause a novel, severe intellectual disability syndrome with cataracts and variable microcephaly. Genome Medicine, 2021, 13, 34.	3.6	18
2768	Complement Factor I Mutation May Contribute to Development of Thrombotic Microangiopathy in Lupus Nephritis. Frontiers in Medicine, 2020, 7, 621609.	1.2	3

#	Article	IF	Citations
2769	Whole Genome Sequencing Prioritizes CHEK2, EWSR1, and TIAM1 as Possible Predisposition Genes for Familial Non-Medullary Thyroid Cancer. Frontiers in Endocrinology, 2021, 12, 600682.	1.5	13
2770	Sequencing of 53,831 diverse genomes from the NHLBI TOPMed Program. Nature, 2021, 590, 290-299.	13.7	1,069
2771	Gain-of-function variants in SYK cause immune dysregulation and systemic inflammation in humans and mice. Nature Genetics, 2021, 53, 500-510.	9.4	56
2772	Common and Rare Genetic Variants That Could Contribute to Severe Otitis Media in an Australian Aboriginal Population. Clinical Infectious Diseases, 2021, 73, 1860-1870.	2.9	4
2774	Next-generation sequencing for inborn errors of immunity. Human Immunology, 2021, 82, 871-882.	1.2	12
2775	Expanding the clinical phenotype of the ultraâ€rare <scp>Skrabanâ€Deardorff</scp> syndrome: Two novel individuals with <scp>WDR26</scp> lossâ€ofâ€function variants and a literature review. American Journal of Medical Genetics, Part A, 2021, 185, 1712-1720.	0.7	6
2776	Identification of known and novel familial cancer genes in Swedish colorectal cancer families. International Journal of Cancer, 2021, 149, 627-634.	2.3	0
2777	Prioritizing non-coding regions based on human genomic constraint and sequence context with deep learning. Nature Communications, 2021, 12, 1504.	5.8	40
2779	A Novel Pathogenic HSPG2 Mutation in Schwartzâ€“Jampel Syndrome. Frontiers in Neurology, 2021, 12, 632336.	1.1	11
2782	Germline and Tumor Sequencing as a Diagnostic Tool To Resolve Suspected Lynch Syndrome. Journal of Molecular Diagnostics, 2021, 23, 358-371.	1.2	12
2783	A Drosophila platform identifies a novel, personalized therapy for a patient with adenoid cystic carcinoma. IScience, 2021, 24, 102212.	1.9	23
2784	Understanding the Origins of Loss of Protein Function by Analyzing the Effects of Thousands of Variants on Activity and Abundance. Molecular Biology and Evolution, 2021, 38, 3235-3246.	3.5	65
2785	Clinical and molecular findings in children and young adults with persistent low alkaline phosphatase concentrations. Annals of Clinical Biochemistry, 2021, 58, 000456322110001.	0.8	2
2786	SLCO1B1: Application and Limitations of Deep Mutational Scanning for Genomic Missense Variant Function. Drug Metabolism and Disposition, 2021, 49, 395-404.	1.7	17
2787	Prioritization of candidate genes for a South African family with Parkinsonâ€™s disease using in-silico tools. PLoS ONE, 2021, 16, e0249324.	1.1	9
2788	Exome-Wide Search for Genes Associated With Central Nervous System Inflammatory Demyelinating Diseases Following CHIKV Infection: The Tip of the Iceberg. Frontiers in Genetics, 2021, 12, 639364.	1.1	8
2790	Autosomal Recessive Rod-Cone Dystrophy Associated With Compound Heterozygous Variants in ARL3 Gene. Frontiers in Cell and Developmental Biology, 2021, 9, 635424.	1.8	18
2791	Evidence of synergism among three genetic variants in a patient with LMNA-related lipodystrophy and amyotrophic lateral sclerosis leading to a remarkable nuclear phenotype. Molecular and Cellular Biochemistry, 2021, 476, 2633-2650.	1.4	4

#	Article	IF	Citations
2792	Remus: A Web Application for Prioritization of Regulatory Regions and Variants in Monogenic Diseases. Frontiers in Genetics, 2021, 12, 638960.	1.1	1
2793	Novel ORAI1 Mutation Disrupts Channel Trafficking Resulting in Combined Immunodeficiency. Journal of Clinical Immunology, 2021, 41, 1004-1015.	2.0	5
2794	Genotypeâ€“Phenotype Relations for the Atypical Parkinsonism Genes: MDSGene Systematic Review. Movement Disorders, 2021, 36, 1499-1510.	2.2	22
2795	Classifying molecular phenotypes of <scp>G6PC</scp> variants for pathogenic properties and to guide therapeutic development. JIMD Reports, 2021, 60, 56-66.	0.7	2
2796	In silico screening and analysis of nonsynonymous SNPs in human CYP1A2 to assess possible associations with pathogenicity and cancer susceptibility. Scientific Reports, 2021, 11, 4977.	1.6	12
2797	A novel missense TGFBI variant p.(Ser591Phe) in a Finnish family with variant lattice corneal dystrophy. European Journal of Ophthalmology, 2021, , 112067212199730.	0.7	1
2798	Novel BTK mutation in X-linked agammaglobulinemia: Report of a 17-year-old male. Allergologia Et Immunopathologia, 2021, 49, 80-83.	1.0	3
2799	Variant Spectrum of Formin Homology 2 Domainâ€Containing 3 Gene in Chinese Patients With Hypertrophic Cardiomyopathy. Journal of the American Heart Association, 2021, 10, e018236.	1.6	10
2800	In silico prediction of HBD gene variants in the Iranian population. Egyptian Journal of Medical Human Genetics, 2021, 22, .	0.5	0
2801	Establishing intellectual disability as the key feature of patients with biallelic <scp>RNPC3</scp> variants. American Journal of Medical Genetics, Part A, 2021, 185, 1836-1840.	0.7	3
2802	From mutation to mechanism: deciphering the molecular function of genetic variants linked to human ageing. Briefings in Functional Genomics, 2022, 21, 13-23.	1.3	4
2803	Analysis workflow to assess de novo genetic variants from human whole-exome sequencing. STAR Protocols, 2021, 2, 100383.	0.5	7
2804	Low Diversity of Human Variation Despite Mostly Mild Functional Impact of De Novo Variants. Frontiers in Molecular Biosciences, 2021, 8, 635382.	1.6	2
2805	Evidence for pathogenicity of variant ATM Val1729Leu in a family with ataxia telangiectasia. Neurogenetics, 2021, 22, 143-147.	0.7	2
2806	An Asian-specific MPL genetic variant alters JAKâ€“STAT signaling and influences platelet count in the population. Human Molecular Genetics, 2021, 30, 836-842.	1.4	4
2808	A Case of Severe Combined Immunodeficiency Missed by Newborn Screening. Journal of Clinical Immunology, 2021, 41, 1352-1355.	2.0	4
2809	Variants in NEB and RIF1 genes on chr2q23 are associated with skeletal muscle index in Koreans: genome-wide association study. Scientific Reports, 2021, 11, 2333.	1.6	1
2810	Activation of cryptic donor splice sites by non-coding and coding PAX6 variants contributes to congenital aniridia. Journal of Medical Genetics, 2021, , jmedgenet-2020-106932.	1.5	8

#	Article	IF	Citations
2811	Identifying the causes of recurrent pregnancy loss in consanguineous couples using whole exome sequencing on the products of miscarriage with no chromosomal abnormalities. Scientific Reports, 2021, 11, 6952.	1.6	15
2812	Profiling chromatin accessibility in pediatric acute lymphoblastic leukemia identifies subtype-specific chromatin landscapes and gene regulatory networks. Leukemia, 2021, 35, 3078-3091.	3.3	15
2813	The Impact of Modern Technologies on Molecular Diagnostic Success Rates, with a Focus on Inherited Retinal Dystrophy and Hearing Loss. International Journal of Molecular Sciences, 2021, 22, 2943.	1.8	6
2814	Genetic and phenotypic analysis of 101 patients with developmental delay or intellectual disability using wholeâ€exome sequencing. Clinical Genetics, 2021, 100, 40-50.	1.0	17
2815	Identification of Deep-Intronic Splice Mutations in a Large Cohort of Patients With Inherited Retinal Diseases. Frontiers in Genetics, 2021, 12, 647400.	1.1	29
2816	The genetic architecture of structural leftâ€“right asymmetry of the human brain. Nature Human Behaviour, 2021, 5, 1226-1239.	6.2	70
2817	A linkage and exome study of multiplex families with bipolar disorder implicates rare coding variants of ANK3 and additional rare alleles at 10q11-q21. Journal of Psychiatry and Neuroscience, 2021, 46, E247-E257.	1.4	6
2818	Contribution of coding/non-coding variants in NUS1 to late-onset sporadic Parkinson's disease. Parkinsonism and Related Disorders, 2021, 84, 29-34.	1.1	2
2819	A New Insight for the Identification of Oncogenic Variants in Breast and Prostate Cancers in Diverse Human Populations, With a Focus on Latinos. Frontiers in Pharmacology, 2021, 12, 630658.	1.6	3
2821	A clinical transcriptome approach to patient stratification and therapy selection in acute myeloid leukemia. Nature Communications, 2021, 12, 2474.	5.8	49
2822	Biallelic hypomorphic variants in ALDH1A2 cause a novel lethal human multiple congenital anomaly syndrome encompassing diaphragmatic, pulmonary, and cardiovascular defects. Human Mutation, 2021, 42, 506-519.	1.1	12
2823	A rare variant in <scp>EZH2</scp> is associated with prostate cancer risk. International Journal of Cancer, 2021, 149, 1089-1099.	2.3	9
2824	Clinical Significance of Variants in the TTN Gene in a Large Cohort of Patients With Sporadic Dilated Cardiomyopathy. Frontiers in Cardiovascular Medicine, 2021, 8, 657689.	1.1	8
2825	Emerging genetic complexity and rare genetic variants in neurodegenerative brain diseases. Genome Medicine, 2021, 13, 59.	3.6	16
2826	usDSM: a novel method for deleterious synonymous mutation prediction using undersampling scheme. Briefings in Bioinformatics, 2021, 22, .	3.2	12
2827	Common Variation in Cytoskeletal Genes Is Associated with Conotruncal Heart Defects. Genes, 2021, 12, 655.	1.0	2
2828	BBS1 branchpoint variant is associated with non-syndromic retinitis pigmentosa. Journal of Medical Genetics, 2022, 59, 438-444.	1.5	13
2829	Confirmation of COL4A6 variants in X-linked nonsyndromic hearing loss and its clinical implications. European Journal of Human Genetics, 2021, , .	1.4	5

#	Article	IF	Citations
2830	Advances in Genomic Discovery and Implications for Personalized Prevention and Medicine: Estonia as Example. Journal of Personalized Medicine, 2021, 11, 358.	1.1	6
2831	Functional assessment of the â€œtwo-hitâ€•model for neurodevelopmental defects in Drosophila and X. laevis. PLoS Genetics, 2021, 17, e1009112.	1.5	12
2832	Familial Idiopathic Basal Ganglia Calcification: A Father-Son Dyad Demonstrate Heterogeneity of Presentation and Disease Progression. Archives of Clinical Neuropsychology, 2022, 37, 217-225.	0.3	0
2833	Using CRISPR to understand and manipulate gene regulation. Development (Cambridge), 2021, 148, .	1.2	9
2835	Variants in STXBP3 are Associated with Very Early Onset Inflammatory Bowel Disease, Bilateral Sensorineural Hearing Loss and Immune Dysregulation. Journal of Crohn's and Colitis, 2021, 15, 1908-1919.	0.6	7
2836	Common and Rare Variant Prediction and Penetrance of IBD in a Large, Multi-ethnic, Health System-based Biobank Cohort. Gastroenterology, 2021, 160, 1546-1557.	0.6	43
2837	Identification of Novel Candidate Genes and Variants for Hearing Loss and Temporal Bone Anomalies. Genes, 2021, 12, 566.	1.0	5
2838	Machine learning-based reclassification of germline variants of unknown significance: The RENOVO algorithm. American Journal of Human Genetics, 2021, 108, 682-695.	2.6	13
2839	Protein design and variant prediction using autoregressive generative models. Nature Communications, 2021, 12, 2403.	5.8	168
2841	Application of Genome Sequencing from Blood to Diagnose Mitochondrial Diseases. Genes, 2021, 12, 607.	1.0	8
2842	Case Report: Compound Heterozygous Variants in MOCS3 Identified in a Chinese Infant With Molybdenum Cofactor Deficiency. Frontiers in Genetics, 2021, 12, 651878.	1.1	5
2843	A Novel Low-Risk Germline Variant in the SH2 Domain of the SRC Gene Affects Multiple Pathways in Familial Colorectal Cancer. Journal of Personalized Medicine, 2021, 11, 262.	1.1	0
2845	Genotype-phenotype correlations of KIF5A stalk domain variants. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2021, 22, 561-570.	1.1	9
2846	Determinants of Response and Intrinsic Resistance to PD-1 Blockade in Microsatellite Instabilityâ€“High Gastric Cancer. Cancer Discovery, 2021, 11, 2168-2185.	7.7	105
2848	<scp>DZIP1</scp> regulates mammalian cardiac valve development through a Cby1â€î²â€catenin mechanism. Developmental Dynamics, 2021, 250, 1432-1449.	0.8	6
2849	Pharmacogenetic Polygenic Risk Score for Bronchodilator Response in Children and Adolescents with Asthma: Proof-of-Concept. Journal of Personalized Medicine, 2021, 11, 319.	1.1	5
2851	Pathogenic missense protein variants affect different functional pathways and proteomic features than healthy population variants. PLoS Biology, 2021, 19, e3001207.	2.6	13
2853	Exome-Wide Association Study on Alanine Aminotransferase Identifies Sequence Variants in the GPAM and APOE Associated With Fatty Liver Disease. Gastroenterology, 2021, 160, 1634-1646.e7.	0.6	82

#	Article	IF	Citations
2854	Biallelic mutations in l-dopachrome tautomerase (DCT) cause infantile nystagmus and oculocutaneous albinism. Human Genetics, 2021, 140, 1157-1168.	1.8	12
2855	Phenotypic and Genetic Spectrum of Autosomal Recessive Bestrophinopathy and Best Vitelliform Macular Dystrophy. , 2021, 62, 22.		11
2856	Incorporation of exomeâ€based CNV analysis makes trioâ€WES a more powerful tool for clinical diagnosis in neurodevelopmental disorders: A retrospective study. Human Mutation, 2021, 42, 990-1004.	1.1	25
2858	Rare proteinâ€coding variants implicate genes involved in risk of suicide death. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2021, 186, 508-520.	1.1	14
2859	Rare Missense Functional Variants at COL4A1 and COL4A2 in Sporadic Intracerebral Hemorrhage. Neurology, 2021, 97, .	1.5	6
2860	Cochlear supporting cells require GAS2 for cytoskeletal architecture and hearing. Developmental Cell, 2021, 56, 1526-1540.e7.	3.1	18
2861	The genetic architecture of the human thalamus and its overlap with ten common brain disorders. Nature Communications, 2021, 12, 2909.	5.8	25
2862	In Silico Predictions of KCNQ Variant Pathogenicity in Epilepsy. Pediatric Neurology, 2021, 118, 48-54.	1.0	2
2863	The causes of Fanconi anemia in South Asia and the Middle East: A case series and review of the literature. Molecular Genetics & Genomic Medicine, 2021, 9, e1693.	0.6	2
2864	Familial Occurrence of Adult Granulosa Cell Tumors: Analysis of Whole-Genome Germline Variants. Cancers, 2021, 13, 2430.	1.7	1
2866	A method to delineate de novo missense variants across pathways prioritizes genes linked to autism. Science Translational Medicine, 2021, 13, .	5.8	19
2867	Single-cell transcriptomics trajectory and molecular convergence of clinically relevant mutations in Brugada syndrome. American Journal of Physiology - Heart and Circulatory Physiology, 2021, 320, H1935-H1948.	1.5	6
2868	Low frequency variants associated with leukocyte telomere length in the Singapore Chinese population. Communications Biology, 2021, 4, 519.	2.0	15
2869	Feasibility of predicting allele specific expression from DNA sequencing using machine learning. Scientific Reports, 2021, 11, 10606.	1.6	4
2870	Identification of missense MAB21L1 variants in microphthalmia and aniridia. Human Mutation, 2021, 42, 877-890.	1.1	13
2871	GIMAP5 maintains liver endothelial cell homeostasis and prevents portal hypertension. Journal of Experimental Medicine, 2021, 218, .	4.2	22
2872	The GBA-370Rec Parkinson's disease risk haplotype harbors a potentially pathogenic variant in the mitochondrial gene SLC25A44. Molecular Genetics and Metabolism, 2021, 133, 109-112.	0.5	2
2873	The DBSAV Database: Predicting Deleteriousness of Single Amino Acid Variations in the Human Proteome. Journal of Molecular Biology, 2021, 433, 166915.	2.0	15

#	Article	IF	Citations
2875	Purifying selection on noncoding deletions of human regulatory loci detected using their cellular pleiotropy. Genome Research, 2021, 31, 935-946.	2.4	5
2877	Impact of rare and common genetic variation in the interleukin-1 pathway on human cytokine responses. Genome Medicine, 2021, 13, 94.	3.6	5
2878	The clinical and imaging features of FLNA positive and negative periventricular nodular heterotopia. Biomedical Journal, 2022, 45, 542-548.	1.4	3
2879	Recurrent <scp>NFIA K125E</scp> substitution represents a lossâ€ofâ€function allele: Sensitive in vitro and in vivo assays forÂnontruncating alleles. American Journal of Medical Genetics, Part A, 2021, 185, 2084-2093.	0.7	9
2880	Dysgenesis and Dysfunction of the Pancreas and Pituitary Due toFOXA2Gene Defects. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e4142-e4154.	1.8	6
2881	Genetic underpinnings of sociability in the general population. Neuropsychopharmacology, 2021, 46, 1627-1634.	2.8	18
2882	GWAS of allometric body-shape indices in UK Biobank identifies loci suggesting associations with morphogenesis, organogenesis, adrenal cell renewal and cancer. Scientific Reports, 2021, 11, 10688.	1.6	68
2883	Functional analysis of PCSK9 3â€²UTR variants and mRNAâ€“miRNA interactions in patients with familial hypercholesterolemia. Epigenomics, 2021, 13, 779-791.	1.0	5
2884	A novel ENAM mutation causes hypoplastic amelogenesis imperfecta. Oral Diseases, 2022, 28, 1610-1619.	1.5	6
2886	Biallelic variants in VPS50 cause a neurodevelopmental disorder with neonatal cholestasis. Brain, 2021, 144, 3036-3049.	3.7	4
2887	Contribution of Evolutionary Selected Immune Gene Polymorphism to Immune-Related Disorders: The Case of Lymphocyte Scavenger Receptors CD5 and CD6. International Journal of Molecular Sciences, 2021, 22, 5315.	1.8	6
2888	Neurodevelopmental phenotypes associated with pathogenic variants in SLC6A1. Journal of Medical Genetics, 2022, 59, 536-543.	1.5	18
2889	Exonic variants undergoing allele-specific selection in cancers. BMC Medical Genomics, 2021, 14, 142.	0.7	0
2890	Focused Strategies for Defining the Genetic Architecture of Congenital Heart Defects. Genes, 2021, 12, 827.	1.0	8
2891	Investigation of genetic loci shared between bipolar disorder and risk-taking propensity: potential implications for pharmacological interventions. Neuropsychopharmacology, 2021, 46, 1680-1692.	2.8	2
2893	Refinement of the clinical variant interpretation framework by statistical evidence and machine learning. Med, 2021, 2, 611-632.e9.	2.2	1
2894	Identifying and elucidating the roles of Y198N and Y204F mutations in the PAH enzyme through molecular dynamic simulations. Journal of Biomolecular Structure and Dynamics, 2021, , 1-12.	2.0	3
2895	The clinical and functional effects of TERT variants in myelodysplastic syndrome. Blood, 2021, 138, 898-911.	0.6	27

#	Article	IF	Citations
2896	Novel TUBA4A Variant Associated With Familial Frontotemporal Dementia. Neurology: Genetics, 2021, 7, e596.	0.9	18
2898	Genetic and immunologic findings in children with recurrent aphthous stomatitis with systemic inflammation. Pediatric Rheumatology, 2021, 19, 70.	0.9	12
2899	A novel LMNA indel mutation identified in a family with atrioventricular block and atrial fibrillation. Medicine (United States), 2021, 100, e25910.	0.4	2
2900	ELASPIC2 (EL2): Combining Contextualized Language Models and Graph Neural Networks to Predict Effects of Mutations. Journal of Molecular Biology, 2021, 433, 166810.	2.0	24
2902	A Recurrent <scp>EIF2AK2</scp> Missense Variant Causes Autosomalâ€Dominant Isolated Dystonia. Annals of Neurology, 2021, 89, 1257-1258.	2.8	10
2903	DeepFun: a deep learning sequence-based model to decipher non-coding variant effect in a tissue- and cell type-specific manner. Nucleic Acids Research, 2021, 49, W131-W139.	6.5	15
2904	OpenAnnotate: a web server to annotate the chromatin accessibility of genomic regions. Nucleic Acids Research, 2021, 49, W483-W490.	6.5	17
2905	Large-scale plasma proteomic analysis identifies proteins and pathways associated with dementia risk. Nature Aging, 2021, 1, 473-489.	5.3	69
2906	A 7-year old female with arthrogryposis multiplex congenita, Duane retraction syndrome, and Marcus Gunn phenomenon due to a ZC4H2 gene mutation: a clinical presentation of the Wieacker-Wolff syndrome. Ophthalmic Genetics, 2021, 42, 612-614.	0.5	5
2907	Mitochondrial genome copy number measured by DNA sequencing in human blood is strongly associated with metabolic traits via cell-type composition differences. Human Genomics, 2021, 15, 34.	1.4	7
2909	Whole-genome sequencing identifies functional noncoding variation in SEMA3C that cosegregates with dyslexia in a multigenerational family. Human Genetics, 2021, 140, 1183-1200.	1.8	5
2910	A Homozygous Deep Intronic Mutation Alters the Splicing of Nebulin Gene in a Patient With Nemaline Myopathy. Frontiers in Neurology, 2021, 12, 660113.	1.1	6
2911	Deep sequencing of 1320 genes reveals the landscape of protein-truncating variants and their contribution to psoriasis in 19,973 Chinese individuals. Genome Research, 2021, 31, 1150-1158.	2.4	5
2912	Apparent but unconfirmed digenism in an Iranian consanguineous family with syndromic Retinal Disease. Experimental Eye Research, 2021, 207, 108533.	1.2	1
2913	Phenotypic diversity, disease progression, and pathogenicity of <scp>MVK</scp> missense variants in mevalonic aciduria. Journal of Inherited Metabolic Disease, 2021, 44, 1272-1287.	1.7	17
2914	A comparison of genotyping arrays. European Journal of Human Genetics, 2021, 29, 1611-1624.	1.4	43
2915	PHKA2 variants expand the phenotype of phosphorylase B kinase deficiency to include patients with ketotic hypoglycemia only. American Journal of Medical Genetics, Part A, 2021, 185, 2959-2975.	0.7	2
2916	Human STAT3 variants underlie autosomal dominant hyper-IgE syndrome by negative dominance. Journal of Experimental Medicine, 2021, 218, .	4.2	30

#	Article	IF	Citations
2917	Genome sequencing unveils a regulatory landscape of platelet reactivity. Nature Communications, 2021, 12, 3626.	5.8	29
2918	Rare variants regulate expression of nearby individual genes in multiple tissues. PLoS Genetics, 2021, 17, e1009596.	1.5	6
2920	Molecular Inversion Probe-Based Sequencing of USH2A Exons and Splice Sites as a Cost-Effective Screening Tool in USH2 and arRP Cases. International Journal of Molecular Sciences, 2021, 22, 6419.	1.8	8
2922	Computational Tools to Assess the Functional Consequences of Rare and Noncoding Pharmacogenetic Variability. Clinical Pharmacology and Therapeutics, 2021, 110, 626-636.	2.3	16
2923	Analysis of highâ€risk pedigrees identifies 11 candidate variants for Alzheimer's disease. Alzheimer's and Dementia, 2022, 18, 307-317.	0.4	10
2924	Shared genetic architecture between neuroticism, coronary artery disease and cardiovascular risk factors. Translational Psychiatry, 2021, 11, 368.	2.4	10
2925	Bioinformatics Accelerates the Major Tetrad: A Real Boost for the Pharmaceutical Industry. International Journal of Molecular Sciences, 2021, 22, 6184.	1.8	16
2926	Co-Occurring Heterozygous CNOT3 and SMAD6 Truncating Variants: Unusual Presentation and Refinement of the IDDSADF Phenotype. Genes, 2021, 12, 1009.	1.0	3
2927	Towards a New, Endophenotype-Based Strategy for Pathogenicity Prediction in BRCA1 and BRCA2: In Silico Modeling of the Outcome of HDR/SGE Assays for Missense Variants. International Journal of Molecular Sciences, 2021, 22, 6226.	1.8	0
2928	Genetic Characterization of Cancer of Unknown Primary Using Liquid Biopsy Approaches. Frontiers in Cell and Developmental Biology, 2021, 9, 666156.	1.8	12
2929	Postmortem detection of COL gene family variants in two aortic dissection cases. International Journal of Legal Medicine, 2022, 136, 85-91.	1.2	2
2930	Disruption of NEUROD2 causes a neurodevelopmental syndrome with autistic features via cell-autonomous defects in forebrain glutamatergic neurons. Molecular Psychiatry, 2021, 26, 6125-6148.	4.1	21
2931	Extending the allelic spectrum at noncoding risk loci of orofacial clefting. Human Mutation, 2021, 42, 1066-1078.	1.1	3
2932	Adult GAMT deficiency: A literature review and report of two siblings. Molecular Genetics and Metabolism Reports, 2021, 27, 100761.	0.4	6
2933	Protective Role of DHEAS in Age-related Changes in Bone Mass and Fracture Risk. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e4580-e4592.	1.8	11
2935	KCNK18 Biallelic Variants Associated with Intellectual Disability and Neurodevelopmental Disorders Alter TRESK Channel Activity. International Journal of Molecular Sciences, 2021, 22, 6064.	1.8	3
2936	Adducted Thumb and Peripheral Polyneuropathy: Diagnostic Supports in Suspecting Whiteâ€"Sutton Syndrome: Case Report and Review of the Literature. Genes, 2021, 12, 950.	1.0	5
2937	Functional characterization of 105 factor H variants associated with aHUS: lessons for variant classification. Blood, 2021, 138, 2185-2201.	0.6	29

#	Article	IF	Citations
2938	Biallelic variants in RNU12 cause CDAGS syndrome. Human Mutation, 2021, 42, 1042-1052.	1.1	5
2939	Genome-wide association analysis of metabolic syndrome quantitative traits in the GENNID multiethnic family study. Diabetology and Metabolic Syndrome, 2021, 13, 59.	1.2	9
2942	Molecular predictors of the outcome of paclitaxel plus carboplatin neoadjuvant therapy in high-grade serous ovarian cancer patients. Cancer Chemotherapy and Pharmacology, 2021, 88, 439-450.	1.1	11
2943	Whole-Genome Sequencing Identifies Two Novel Rare Mutations in BMP5 and BMP2 in Monozygotic Twins With Microtia. Journal of Craniofacial Surgery, 2022, 33, e212-e217.	0.3	6
2944	A semi-supervised deep learning approach for predicting the functional effects of genomic non-coding variations. BMC Bioinformatics, 2021, 22, 128.	1.2	4
2945	APP, PSEN1, and PSEN2 Variants in Alzheimerâ€™s Disease: Systematic Re-evaluation According to ACMG Guidelines. Frontiers in Aging Neuroscience, 2021, 13, 695808.	1.7	33
2946	Impact of genetic tests on survivors of paediatric sudden cardiac arrest. Archives of Disease in Childhood, 2021, , archdischild-2020-321532.	1.0	0
2947	Assessment of the gene mosaicism burden in blood and its implications for immune disorders. Scientific Reports, 2021, 11, 12940.	1.6	5
2948	Heterozygous OAS1 gain-of-function variants cause an autoinflammatory immunodeficiency. Science Immunology, 2021, 6, .	5.6	36
2949	PIGG variant pathogenicity assessment reveals characteristic features within 19 families. Genetics in Medicine, 2021, 23, 1873-1881.	1.1	5
2950	How Machine Learning and Statistical Models Advance Molecular Diagnostics of Rare Disorders Via Analysis of RNA Sequencing Data. Frontiers in Molecular Biosciences, 2021, 8, 647277.	1.6	12
2951	A computational approach for detecting physiological homogeneity in the midst of genetic heterogeneity. American Journal of Human Genetics, 2021, 108, 1012-1025.	2.6	6
2952	Proximal variants in <scp>CCND2</scp> associated with microcephaly, short stature, and developmental delay: A case series and review of inverse brain growth phenotypes. American Journal of Medical Genetics, Part A, 2021, 185, 2719-2738.	0.7	14
2953	CFH and CFHR Copy Number Variations in C3 Glomerulopathy and Immune Complex-Mediated Membranoproliferative Glomerulonephritis. Frontiers in Genetics, 2021, 12, 670727.	1.1	11
2954	Novel loci and potential mechanisms of major depressive disorder, bipolar disorder, and schizophrenia. Science China Life Sciences, 2022, 65, 167-183.	2.3	9
2955	A Curriculum for Genomic Education of Molecular Genetic Pathology Fellows. Journal of Molecular Diagnostics, 2021, 23, 1218-1240.	1.2	4
2956	De novo and bi-allelic variants in AP1G1 cause neurodevelopmental disorder with developmental delay, intellectual disability, and epilepsy. American Journal of Human Genetics, 2021, 108, 1330-1341.	2.6	18
2957	Multiple Acyl-CoA Dehydrogenase Deficiency with Variable Presentation Due to a Homozygous Mutation in a Bedouin Tribe. Genes, 2021, 12, 1140.	1.0	1

#	Article	IF	Citations
2958	An artificial neural network approach integrating plasma proteomics and genetic data identifies PLXNA4 as a new susceptibility locus for pulmonary embolism. Scientific Reports, 2021, 11, 14015.	1.6	8
2959	Clinical and genetic risk factors for radiationâ€associated ototoxicity: A report from the Childhood Cancer Survivor Study and the St. Jude Lifetime Cohort. Cancer, 2021, 127, 4091-4102.	2.0	6
2962	Exome variant discrepancies due to reference-genome differences. American Journal of Human Genetics, 2021, 108, 1239-1250.	2.6	36
2964	A machine learning approach to brain epigenetic analysis reveals kinases associated with Alzheimerâ€™s disease. Nature Communications, 2021, 12, 4472.	5.8	28
2965	Genes affecting ionizing radiation survival identified through combined exome sequencing and functional screening. Human Mutation, 2021, 42, 1124-1138.	1.1	0
2967	Myopia in African Americans Is Significantly Linked to Chromosome 7p15.2-14.2. , 2021, 62, 16.		2
2968	Artificial Intelligence in Chemistry: Current Trends and Future Directions. Journal of Chemical Information and Modeling, 2021, 61, 3197-3212.	2.5	80
2969	Sequencing for germline mutations in Swedish breast cancer families reveals novel breast cancer risk genes. Scientific Reports, 2021, 11, 14737.	1.6	2
2970	Genetic analysis of four consanguineous multiplex families with inflammatory bowel disease. Gastroenterology Report, 2021, 9, 521-532.	0.6	5
2971	REVA as A Well-curated Database for Human Expression-modulating Variants. Genomics, Proteomics and Bioinformatics, 2021, 19, 590-601.	3.0	0
2973	Mutation analysis of SOD1, C9orf72, TARDBP and FUS genes in ethnically-diverse Malaysian patients with amyotrophic lateral sclerosis (ALS). Neurobiology of Aging, 2021, 108, 200-206.	1.5	7
2974	Human autoinflammatory disease reveals ELF4 as a transcriptional regulator of inflammation. Nature Immunology, 2021, 22, 1118-1126.	7.0	30
2975	Recent ultra-rare inherited variants implicate new autism candidate risk genes. Nature Genetics, 2021, 53, 1125-1134.	9.4	68
2976	Humans with inherited TÂcell CD28 deficiency are susceptible to skin papillomaviruses but are otherwise healthy. Cell, 2021, 184, 3812-3828.e30.	13.5	53
2977	MRIâ€Based Iron Phenotyping and Patient Selection for Nextâ€Generation Sequencing of Nonâ€“Homeostatic Iron Regulator Hemochromatosis Genes. Hepatology, 2021, 74, 2424-2435.	3.6	8
2979	SCN1A-related epilepsy with recessive inheritance: Two further families. European Journal of Paediatric Neurology, 2021, 33, 121-124.	0.7	4
2980	Unveiling the genetic variation of severe continuous/mixed-type ossification of the posterior longitudinal ligament by whole-exome sequencing and bioinformatic analysis. Spine Journal, 2021, 21, 1847-1856.	0.6	5
2981	Quantifying Intensities of Transcription Factor-DNA Binding by Learning From an Ensemble of Protein Binding Microarrays. IEEE Journal of Biomedical and Health Informatics, 2021, 25, 2811-2819.	3.9	2

#	Article	IF	Citations
2982	The role of CDHR3 in susceptibility to otitis media. Journal of Molecular Medicine, 2021, 99, 1571-1583.	1.7	4
2983	Atypical pantothenate kinase-associated neurodegeneration with variable phenotypes in an Egyptian family. Heliyon, 2021, 7, e07469.	1.4	0
2984	Human AGR2 Deficiency Causes Mucus Barrier Dysfunction and Infantile Inflammatory Bowel Disease. Cellular and Molecular Gastroenterology and Hepatology, 2021, 12, 1809-1830.	2.3	26
2985	A systematic analysis of protein-altering exonic variants in chronic obstructive pulmonary disease. American Journal of Physiology - Lung Cellular and Molecular Physiology, 2021, 321, L130-L143.	1.3	11
2986	Efficacy of computational predictions of the functional effect of idiosyncratic pharmacogenetic variants. PeerJ, 2021, 9, e11774.	0.9	2
2987	Genetic prediction of complex traits with polygenic scores: a statistical review. Trends in Genetics, 2021, 37, 995-1011.	2.9	55
2989	Clinical characteristics and genetic spectrum of 26 individuals of Chinese origin with primary ciliary dyskinesia. Orphanet Journal of Rare Diseases, 2021, 16, 293.	1.2	11
2990	A novel homozygous RAG1 mutation is associated with severe combined immunodeficiency and neurological presentations. Allergologia Et Immunopathologia, 2021, 49, 91-97.	1.0	1
2992	Re-analysis of whole-exome sequencing data reveals a novel splicing variant in the SLC2A1 in a patient with GLUT1 Deficiency Syndrome 1 accompanied by hemangioma: a case report. BMC Medical Genomics, 2021, 14, 197.	0.7	1
2993	Variants in LSM7 impair LSM complexes assembly, neurodevelopment in zebrafish and may be associated with an ultra-rare neurological disease. Human Genetics and Genomics Advances, 2021, 2, 100034.	1.0	3
2994	A Novel Pathogenic NOD2 Variant in a Mother and Daughter with Blau Syndrome. Ophthalmic Genetics, 2021, 42, 1-12.	0.5	3
2995	Impact of Genetic Variation in Gene Regulatory Sequences: A Population Genomics Perspective. Frontiers in Genetics, 2021, 12, 660899.	1.1	2
2996	Clinical evaluation of RB1 genetic testing reveals novel mutations in Vietnamese patients with retinoblastoma. Molecular and Clinical Oncology, 2021, 15, 182.	0.4	2
2997	Exome sequencing reveals novel rare variants in Iranian familial multiple sclerosis: The importance of POLD2 in the disease pathogenesis. Genomics, 2021, 113, 2645-2655.	1.3	5
2998	Phenotypic variability of a TREX1 variant in Aicardi-Goutieres type 1 patients from the Indian subcontinent. European Journal of Medical Genetics, 2021, 64, 104291.	0.7	1
2999	TUMOSPEC: A Nation-Wide Study of Hereditary Breast and Ovarian Cancer Families with a Predicted Pathogenic Variant Identified through Multigene Panel Testing. Cancers, 2021, 13, 3659.	1.7	4
3000	Accelerated expansion of pathogenic mitochondrial DNA heteroplasmies in Huntingtonâ€™s disease. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	15
3001	Extensive bidirectional genetic overlap between bipolar disorder and cardiovascular disease phenotypes. Translational Psychiatry, 2021, 11, 407.	2.4	16

#	Article	IF	Citations
3002	Novel FGFR1 Variants Are Associated with Congenital Scoliosis. Genes, 2021, 12, 1126.	1.0	2
3003	Shifting landscapes of human MTHFR missense-variant effects. American Journal of Human Genetics, 2021, 108, 1283-1300.	2.6	33
3004	Germline Alterations in Patients With IBD-associated Colorectal Cancer. Inflammatory Bowel Diseases, 2022, 28, 447-454.	0.9	6
3005	GFAP variants leading to infantile Alexander disease: Phenotype and genotype analysis of 135 cases and report of a de novo variant. Clinical Neurology and Neurosurgery, 2021, 207, 106754.	0.6	3
3006	Machine learning applications for therapeutic tasks with genomics data. Patterns, 2021, 2, 100328.	3.1	14
3007	Frequency of Heterozygous Parkin (PRKN) Variants and Penetrance of Parkinson's Disease Risk Markers in the Population-Based CHRIS Cohort. Frontiers in Neurology, 2021, 12, 706145.	1.1	14
3008	Novel compound heterozygous missense mutations in GDAP1 cause Charcotâ€“Marieâ€“Tooth type 4A. Journal of Genetics, 2021, 100, 1.	0.4	0
3009	Uniting biobank resources reveals novel genetic pathways modulating susceptibility for atopic dermatitis. Journal of Allergy and Clinical Immunology, 2022, 149, 1105-1112.e9.	1.5	41
3010	Family-based exome sequencing identifies RBM45 as a possible candidate gene for frontotemporal dementia and amyotrophic lateral sclerosis. Neurobiology of Disease, 2021, 156, 105421.	2.1	2
3011	An Overview of the Genetics of ABCA4 Retinopathies, an Evolving Story. Genes, 2021, 12, 1241.	1.0	22
3012	Homozygous missense mutation in UQCRC2 associated with severe encephalomyopathy, mitochondrial complex III assembly defect and activation of mitochondrial protein quality control. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2021, 1867, 166147.	1.8	11
3013	Human TBK1 deficiency leads to autoinflammation driven by TNF-induced cell death. Cell, 2021, 184, 4447-4463.e20.	13.5	64
3015	Exome sequencing in children with clinically suspected <scp>maturityâ€onset</scp> diabetes of the young. Pediatric Diabetes, 2021, 22, 960-968.	1.2	6
3016	Genetic Analysis of a Cohort of 275 Patients with Hyper-IgE Syndromes and/or Chronic Mucocutaneous Candidiasis. Journal of Clinical Immunology, 2021, 41, 1804-1838.	2.0	12
3017	Interacting evolutionary pressures drive mutation dynamics and health outcomes in aging blood. Nature Communications, 2021, 12, 4921.	5.8	11
3018	Allele-specific RT-PCR for the rapid detection of recurrent SLC12A3 mutations for Gitelman syndrome. Npj Genomic Medicine, 2021, 6, 68.	1.7	2
3019	PNPT1, MYO15A, PTPRQ, and SLC12A2â€associated genetic and phenotypic heterogeneity among hearing impaired assortative mating families in Southern India. Annals of Human Genetics, 2022, 86, 1-13.	0.3	5
3020	Assigning function to SNPs: Considerations when interpreting genetic variation. Seminars in Cell and Developmental Biology, 2022, 121, 135-142.	2.3	13

#	Article	IF	Citations
3021	Mutation burden in patients with small unrepaired atrial septal defects. International Journal of Cardiology Congenital Heart Disease, 2021, 4, 100164.	0.2	6
3022	Genetic analysis of dietary intake identifies new loci and functional links with metabolic traits. Nature Human Behaviour, 2022, 6, 155-163.	6.2	22
3025	A girl with a mutation of the ciliary gene CC2D2A presenting with FSGS and nephronophthisis. CEN Case Reports, 2021, , 1.	0.5	5
3026	X-CNV: genome-wide prediction of the pathogenicity of copy number variations. Genome Medicine, 2021, 13, 132.	3.6	24
3027	A domain damage index to prioritizing the pathogenicity of missense variants. Human Mutation, 2021, 42, 1503-1517.	1.1	0
3028	DeepnsSNPs: Accurate prediction of non-synonymous single-nucleotide polymorphisms by combining multi-scale convolutional neural network and residue environment information. Chemometrics and Intelligent Laboratory Systems, 2021, 215, 104326.	1.8	10
3029	Abatacept for treatment-refractory pediatric CTLA4-haploinsufficiency. Clinical Immunology, 2021, 229, 108779.	1.4	18
3030	Phenotypes and genotypes in nonâ€consanguineous and consanguineous primary microcephaly: High incidence of epilepsy. Molecular Genetics & Genomic Medicine, 2021, 9, e1768.	0.6	6
3031	Somatic genetic rescue of a germline ribosome assembly defect. Nature Communications, 2021, 12, 5044.	5.8	44
3032	De novo ARF3 variants cause neurodevelopmental disorder with brain abnormality. Human Molecular Genetics, 2021, 31, 69-81.	1.4	12
3033	Decoding disease: from genomes to networks to phenotypes. Nature Reviews Genetics, 2021, 22, 774-790.	7.7	46
3034	Investigation of the role of matrix metalloproteinases in the genetic etiology of Alzheimer's disease. Neurobiology of Aging, 2021, 104, 105.e1-105.e6.	1.5	8
3035	Integrative genetic, genomic and transcriptomic analysis of heat shock protein and nuclear hormone receptor gene associations with spontaneous preterm birth. Scientific Reports, 2021, 11, 17115.	1.6	12
3039	Exploring the missing heritability in subjects with hearing loss, enlarged vestibular aqueducts, and a single or no pathogenic SLC26A4 variant. Human Genetics, 2022, 141, 465-484.	1.8	3
3040	Rare Germline Variants in ATM Predispose to Prostate Cancer: A PRACTICAL Consortium Study. European Urology Oncology, 2021, 4, 570-579.	2.6	38
3041	Monogenic Diabetes in Youth With Presumed Type 2 Diabetes: Results From the Progress in Diabetes Genetics in Youth (ProDiGY) Collaboration. Diabetes Care, 2021, 44, 2312-2319.	4.3	21
3042	Predicting functional consequences of mutations using molecular interaction network features. Human Genetics, 2022, 141, 1195-1210.	1.8	9
3043	Integrative approach to interpret DYRK1A variants, leading to a frequent neurodevelopmental disorder. Genetics in Medicine, 2021, 23, 2150-2159.	1.1	21

#	Article	IF	Citations
3044	Mapping gene and gene pathways associated with coronary artery disease: a CARDIoGRAM exome and multi-ancestry UK biobank analysis. Scientific Reports, 2021, 11, 16461.	1.6	4
3045	Childhood-onset dystonia-causing KMT2B variants result in a distinctive genomic hypermethylation profile. Clinical Epigenetics, 2021, 13, 157.	1.8	22
3046	Novel genetic variants associated with mortality after unrelated donor allogeneic hematopoietic cell transplantation. EClinicalMedicine, 2021, 40, 101093.	3.2	8
3047	Nonsense-mediated decay is highly stable across individuals and tissues. American Journal of Human Genetics, 2021, 108, 1401-1408.	2.6	15
3049	ZSWIM7Is Associated With Human Female Meiosis and Familial Primary Ovarian Insufficiency. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e254-e263.	1.8	13
3051	Phenotypic and immune functional profiling of patients with suspected Mendelian Susceptibility to Mycobacterial Disease in South Africa. BMC Immunology, 2021, 22, 62.	0.9	4
3052	Improved pathogenicity prediction for rare human missense variants. American Journal of Human Genetics, 2021, 108, 1891-1906.	2.6	51
3053	RAD50 Loss of Function Variants in the Zinc Hook Domain Associated with Higher Risk of Familial Esophageal Squamous Cell Carcinoma. Cancers, 2021, 13, 4715.	1.7	1
3054	A Rare Case of Brachyolmia with Amelogenesis Imperfecta Caused by a New Pathogenic Splicing Variant in LTBP3. Genes, 2021, 12, 1406.	1.0	2
3055	Yield of clinically reportable genetic variants in unselected cerebral palsy by whole genome sequencing. Npj Genomic Medicine, 2021, 6, 74.	1.7	16
3056	Investigating the role ofEGF-CFCgene family in recurrent pregnancy loss through bioinformatics and molecular approaches. Systems Biology in Reproductive Medicine, 2021, 67, 450-462.	1.0	1
3057	Interpretable prioritization of splice variants in diagnostic next-generation sequencing. American Journal of Human Genetics, 2021, 108, 1564-1577.	2.6	36
3058	Whole exome sequencing and functional characterization increase diagnostic yield in siblings with a 46, XY difference of sexual development (DSD). Journal of Steroid Biochemistry and Molecular Biology, 2021, 212, 105908.	1.2	1
3059	Clinical Impact of Detecting Low-Frequency Variants in Cell-Free DNA on Treatment of Castration-Resistant Prostate Cancer. Clinical Cancer Research, 2021, 27, 6164-6173.	3.2	10
3060	PRDM12 Is Transcriptionally Active and Required for Nociceptor Function Throughout Life. Frontiers in Molecular Neuroscience, 2021, 14, 720973.	1.4	7
3061	Inherited human c-Rel deficiency disrupts myeloid and lymphoid immunity to multiple infectious agents. Journal of Clinical Investigation, 2021, 131, .	3.9	21
3063	Phenotype, genotype and long-term prognosis of 40 Chinese patients with isobutyryl-CoA dehydrogenase deficiency and a review of variant spectra in ACAD8. Orphanet Journal of Rare Diseases, 2021, 16, 392.	1.2	8
3064	Developmental genetics of color pattern establishment in cats. Nature Communications, 2021, 12, 5127.	5.8	19

#	Article	IF	Citations
3066	Systematic Evaluation of DNA Sequence Variations on in vivo Transcription Factor Binding Affinity. Frontiers in Genetics, 2021, 12, 667866.	1.1	3
3067	Biochemically deleterious human NFKB1 variants underlie an autosomal dominant form of common variable immunodeficiency. Journal of Experimental Medicine, 2021, 218, .	4.2	32
3068	Genomeâ€wide analysis reveals genetic overlap between alcohol use behaviours, schizophrenia and bipolar disorder and identifies novel shared risk loci. Addiction, 2022, 117, 600-610.	1.7	16
3069	Novel Mutations Detection with Next-Generation Sequencing and Its Association with Clinical Outcome in Unilateral Primary Aldosteronism. Biomedicines, 2021, 9, 1167.	1.4	8
3070	Developmental and temporal characteristics of clonal sperm mosaicism. Cell, 2021, 184, 4772-4783.e15.	13.5	27
3071	Contribution of rare variant associations to neurodegenerative disease presentation. Npj Genomic Medicine, 2021, 6, 80.	1.7	14
3072	Adapting the ACMG/AMP variant classification framework: A perspective from the ClinGen Hemoglobinopathy Variant Curation Expert Panel. Human Mutation, 2022, 43, 1089-1096.	1.1	20
3073	Noncoding sequence variants define a novel regulatory element in the first intron of the N â€acetylglutamate synthase gene. Human Mutation, 2021, 42, 1624-1636.	1.1	3
3075	Elucidating the molecular mechanisms associated with TARS2-related mitochondrial disease. Human Molecular Genetics, 2022, 31, 523-534.	1.4	12
3076	Identifying digenic disease genes via machine learning in the Undiagnosed Diseases Network. American Journal of Human Genetics, 2021, 108, 1946-1963.	2.6	25
3077	Functional Assessment of Missense Variants in the ABCC6 Gene Implicated in Pseudoxanthoma Elasticum, a Heritable Ectopic Mineralization Disorder. Journal of Investigative Dermatology, 2022, 142, 1085-1093.	0.3	2
3079	Rare and de novo variants in 827 congenital diaphragmatic hernia probands implicate LONP1 as candidate risk gene. American Journal of Human Genetics, 2021, 108, 1964-1980.	2.6	22
3080	PRPH2 mutation update: In silico assessment of 245 reported and 7 novel variants in patients with retinal disease. Human Mutation, 2021, 42, 1521-1547.	1.1	13
3081	Whole Exome Sequencing in Individuals with Idiopathic Clubfoot Reveals a Recurrent Filamin B (FLNB) Deletion. Clinical Orthopaedics and Related Research, 2021, Publish Ahead of Print, .	0.7	3
3082	Computational analysis of missense variants in MMP2 gene linked with Winchester syndrome and Nodulosis-Arthropathy-Osteolysis reveals structural shift in protein-protein and protein-ligand complexes. Meta Gene, 2021, 29, 100931.	0.3	2
3084	Genetic Association Between Schizophrenia and Cortical Brain Surface Area and Thickness. JAMA Psychiatry, 2021, 78, 1020.	6.0	43
3085	Genetic Variant Score and Arrhythmogenic Right Ventricular Cardiomyopathy Phenotype in Plakophilin-2 Mutation Carriers. Cardiology, 2021, 146, 763-771.	0.6	3
3086	Exome Sequencing of 5 Families with Severe Early-Onset Periodontitis. Journal of Dental Research, 2022, 101, 151-157.	2.5	5

#	Article	IF	Citations
3087	Deleterious Rare Desmosomal Variants Contribute to Hypertrophic Cardiomyopathy and Are Associated With Distinctive Clinical Features. Canadian Journal of Cardiology, 2022, 38, 41-48.	0.8	6
3088	Frequency and management of medically actionable incidental findings from genome and exome sequencing data: a systematic review. Physiological Genomics, 2021, 53, 373-384.	1.0	11
3089	Somatic reversion impacts myelodysplastic syndromes and acute myeloid leukemia evolution in the short telomere disorders. Journal of Clinical Investigation, 2021, 131, .	3.9	33
3090	VannoPortal: multiscale functional annotation of human genetic variants for interrogating molecular mechanism of traits and diseases. Nucleic Acids Research, 2022, 50, D1408-D1416.	6.5	31
3091	Chromosome X-wide Analysis of Positive Selection in Human Populations: Common and Private Signals of Selection and its Impact on Inactivated Genes and Enhancers. Frontiers in Genetics, 2021, 12, 714491.	1.1	2
3092	A novel variant in DYNC1H1 could contribute to human amyotrophic lateral sclerosis-frontotemporal dementia spectrum.. Journal of Physical Education and Sports Management, 2021, , mcs.a006096.	0.5	8
3094	A guide to machine learning for biologists. Nature Reviews Molecular Cell Biology, 2022, 23, 40-55.	16.1	626
3095	Identification of novel genes for triple-negative breast cancer with semiparametric gene-based analysis. Journal of Applied Statistics, 0, , 1-12.	0.6	1
3096	Dominant <scp>KPNA3</scp> Mutations Cause Infantileâ€Onset Hereditary Spastic Paraplegia. Annals of Neurology, 2021, 90, 738-750.	2.8	5
3097	ECNet is an evolutionary context-integrated deep learning framework for protein engineering. Nature Communications, 2021, 12, 5743.	5.8	66
3099	Molecular and genetic characterization of a large Brazilian cohort presenting hearing loss. Human Genetics, 2022, 141, 519-538.	1.8	6
3100	The prognosis of epilepsy patients with CACNA1H missense variants: A longitudinal cohort study. Seizure: the Journal of the British Epilepsy Association, 2021, 91, 52-59.	0.9	3
3101	Clinical manifestation of CDKL5 deficiency disorder and identified mutations in a cohort of Slovak patients. Epilepsy Research, 2021, 176, 106699.	0.8	1
3102	Characterization of a mutated KCNJ5 gene, G387R, in unilateral primary aldosteronism. Journal of Molecular Endocrinology, 2021, 67, 203-215.	1.1	1
3103	Missense variant in insulin receptor (Y1355H) segregates in family with fatty liver disease. Molecular Metabolism, 2021, 53, 101299.	3.0	1
3104	A rare large duplication of MLH1 identified in Lynch syndrome. Hereditary Cancer in Clinical Practice, 2021, 19, 10.	0.6	2
3105	Artificial intelligence in clinical decision-making for diagnosis of cardiovascular disease using epigenetics mechanisms. , 2021, , 327-345.		2
3106	A catalogue of 863 Rett-syndrome-causing MECP2 mutations and lessons learned from data integration. Scientific Data, 2021, 8, 10.	2.4	12

#	Article	IF	Citations
3108	Advances and challenges in quantitative delineation of the genetic architecture of complex traits. Quantitative Biology, 2021, 9, 168-184.	0.3	0
3109	Achilles Tendon Thickness Assessed by X-ray Predicting a Pathogenic Mutation in Familial Hypercholesterolemia Gene. Journal of Atherosclerosis and Thrombosis, 2022, 29, 816-824.	0.9	20
3110	dbMCS: A Database for Exploring the Mutation Markers of Anti-Cancer Drug Sensitivity. IEEE Journal of Biomedical and Health Informatics, 2021, 25, 4229-4237.	3.9	3
3111	A biallelic variant in CLRN2 causes non-syndromic hearing loss in humans. Human Genetics, 2021, 140, 915-931.	1.8	16
3112	Whole-Exome Sequencing Uncovers Novel Causative Variants and Additional Findings in Three Patients Affected by Glycogen Storage Disease Type VI and Fanconiâ˜Bickel Syndrome. Frontiers in Genetics, 2020, 11, 601566.	1.1	7
3113	Effects of Remimazolam and Propofol on Ca2+ Regulation by Ryanodine Receptor 1 with Malignant Hyperthermia Mutation. BioMed Research International, 2021, 2021, 1-11.	0.9	10
3114	Current cancer driver variant predictors learn to recognize driver genes instead of functional variants. BMC Biology, 2021, 19, 3.	1.7	14
3115	Whole Exome for the Identification of Mutations in CD8+ T-Cells. Methods in Molecular Biology, 2021, 2325, 155-182.	0.4	0
3116	Clinically relevant variants in a large cohort of Indian patients with Marfan syndrome and related disorders identified by next-generation sequencing. Scientific Reports, 2021, 11, 764.	1.6	7
3118	Machine learning in cardiovascular genomics, proteomics, and drug discovery. , 2021, , 325-352.		1
3119	Spectrum of Germline Mutations Within Fanconi Anemiaâ€“Associated Genes Across Populations of Varying Ancestry. JNCI Cancer Spectrum, 2021, 5, .	1.4	3
3121	Polygenic overlap and shared genetic loci between loneliness, severe mental disorders, and cardiovascular disease risk factors suggest shared molecular mechanisms. Translational Psychiatry, 2021, 11, 3.	2.4	29
3122	Pleiotropic genetic influence on birth weight and childhood obesity. Scientific Reports, 2021, 11, 48.	1.6	10
3123	Genomic sequencing of rare diseases. , 2021, , 61-95.		6
3125	The contribution of X-linked coding variation to severe developmental disorders. Nature Communications, 2021, 12, 627.	5.8	33
3126	Long tracks of homozygosity predict the severity of alcohol use disorders in an American Indian population. Molecular Psychiatry, 2021, 26, 2200-2211.	4.1	2
3128	Wide spectrum of NR5A1â€related phenotypes in 46,XY and 46,XX individuals. Birth Defects Research Part C: Embryo Today Reviews, 2016, 108, 309-320.	3.6	76
3129	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. Human Mutation, 2018, 39, 1246-1261.	1.1	31

#	Article	IF	Citations
3130	Next Generation Sequencing (NGS) Strategies for Genetic Testing of Cerebral Cavernous Malformation (CCM) Disease. Methods in Molecular Biology, 2020, 2152, 59-75.	0.4	2
3131	Functional Annotation of Rare Genetic Variants. , 2015, , 57-70.		2
3132	Finding a Needle in a Haystack: Variant Effect Predictor (VEP) Prioritizes Disease Causative Variants from Millions of Neutral Ones. , 2019, , 85-104.		1
3133	Genomic Instability of iPSCs and Challenges in Their Clinical Applications. Advances in Experimental Medicine and Biology, 2019, 1201, 23-47.	0.8	40
3134	Future Paradigm of Breast Cancer Resistance and Treatment. Resistance To Targeted Anti-cancer Therapeutics, 2017, , 155-178.	0.1	2
3135	Adult-onset ataxia or developmental disorder with seizures: two sides of missense changes in CACNA1A. Journal of Neurology, 2017, 264, 1520-1522.	1.8	9
3136	An Integrated Deep-Mutational-Scanning Approach Provides Clinical Insights on PTEN Genotype-Phenotype Relationships. American Journal of Human Genetics, 2020, 106, 818-829.	2.6	38
3137	Genome-wide Study Identifies Association between HLA-Bâˆ—55:01 and Self-Reported Penicillin Allergy. American Journal of Human Genetics, 2020, 107, 612-621.	2.6	34
3138	Novel polymorphisms associated with hyperalphalipoproteinemia and apparent cardioprotection. Journal of Clinical Lipidology, 2018, 12, 110-115.	0.6	8
3139	Untangling a complex web: Computational analyses of tumor molecular profiles to decode driver mechanisms. Journal of Genetics and Genomics, 2020, 47, 595-609.	1.7	5
3140	Comparison of Pathogenicity Prediction Tools on Somatic Variants. Journal of Molecular Diagnostics, 2020, 22, 1383-1392.	1.2	19
3141	A Novel Locus and Candidate Gene for Familial Developmental Dyslexia on Chromosome 4q. Zeitschrift FÃœr Kinder- Und Jugendpsychiatrie Und Psychotherapie, 2020, 48, 478-489.	0.4	5
3142	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nature Communications, 2020, 11, 595.	5.8	35
3143	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	5.8	105
3144	Identification of cancer driver genes based on nucleotide context. Nature Genetics, 2020, 52, 208-218.	9.4	170
3145	A novel de novo variant of GABRA1 causes increased sensitivity for GABA in vitro. Scientific Reports, 2020, 10, 2379.	1.6	18
3146	iFish: predicting the pathogenicity of human nonsynonymous variants using gene-specific/family-specific attributes and classifiers. , 0, .		1
3147	Structural insights into G domain dimerization and pathogenic mutation of OPA1. Journal of Cell Biology, 2020, 219, .	2.3	25

#	Article	IF	Citations
3148	DeMaSk: a deep mutational scanning substitution matrix and its use for variant impact prediction. Bioinformatics, 2021, 36, 5322-5329.	1.8	24
3149	svMIL: predicting the pathogenic effect of TAD boundary-disrupting somatic structural variants throughÂmultiple instance learning. Bioinformatics, 2020, 36, i692-i699.	1.8	4
3150	OUP accepted manuscript. Brain, 2018, 141, e66.	3.7	7
3151	Polygenic Risk Score of Longevity Predicts Longer Survival Across an Age Continuum. Journals of Gerontology - Series A Biological Sciences and Medical Sciences, 2021, 76, 750-759.	1.7	20
3152	A review of innate and adaptive immunity to coccidioidomycosis. Medical Mycology, 2019, 57, S85-S92.	0.3	39
3153	AI-Driver: an ensemble method for identifying driver mutations in personal cancer genomes. NAR Genomics and Bioinformatics, 2020, 2, lqaa084.	1.5	19
3154	Whole exome sequencing in unclassified autoinflammatory diseases: more monogenic diseases in the pipeline?. Rheumatology, 2021, 60, 607-616.	0.9	13
3155	Cholestasis Due to USP53 Deficiency. Journal of Pediatric Gastroenterology and Nutrition, 2021, 72, 667-673.	0.9	18
3319	Mutational landscape of severe combined immunodeficiency patients from Turkey. International Journal of Immunogenetics, 2020, 47, 529-538.	0.8	14
3320	Investigating the genetic susceptibility to exertional heat illness. Journal of Medical Genetics, 2020, 57, 531-541.	1.5	24
3321	Bi-allelic TTC5 variants cause delayed developmental milestones and intellectual disability. Journal of Medical Genetics, 2021, 58, 237-246.	1.5	4
3323	Sialic acid catabolism by N-acetylneuraminate pyruvate lyase is essential for muscle function. JCI Insight, 2018, 3, .	2.3	36
3324	Multiomics dissection of molecular regulatory mechanisms underlying autoimmune-associated noncoding SNPs. JCI Insight, 2020, 5, .	2.3	13
3325	MPEG1/perforin-2 mutations in human pulmonary nontuberculous mycobacterial infections. JCI Insight, 2017, 2, .	2.3	30
3326	Extrapulmonary Aspergillus infection in patients with CARD9 deficiency. JCI Insight, 2016, 1, e89890.	2.3	141
3327	IRS2 mutations linked to invasion in pleomorphic invasive lobular carcinoma. JCI Insight, 2018, 3, .	2.3	18
3328	A nonhuman primate model of inherited retinal disease. Journal of Clinical Investigation, 2019, 129, 863-874.	3.9	78
3329	Rescue of recurrent deep intronic mutation underlying cell typeâ€“dependent quantitative NEMO deficiency. Journal of Clinical Investigation, 2018, 129, 583-597.	3.9	38

#	Article	IF	Citations
3330	Ankyrin-B dysfunction predisposes to arrhythmogenic cardiomyopathy and is amenable to therapy. Journal of Clinical Investigation, 2019, 129, 3171-3184.	3.9	42
3331	Aberrant splicing contributes to severe Î±-spectrinâ€“linked congenital hemolytic anemia. Journal of Clinical Investigation, 2019, 129, 2878-2887.	3.9	24
3332	Polymerase Î´ deficiency causes syndromic immunodeficiency with replicative stress. Journal of Clinical Investigation, 2019, 129, 4194-4206.	3.9	41
3333	Human NK cell deficiency as a result of biallelic mutations in MCM10. Journal of Clinical Investigation, 2020, 130, 5272-5286.	3.9	44
3334	Inherited human IFN-Î³ deficiency underlies mycobacterial disease. Journal of Clinical Investigation, 2020, 130, 3158-3171.	3.9	89
3335	Human CRY1 variants associate with attention deficit/hyperactivity disorder. Journal of Clinical Investigation, 2020, 130, 3885-3900.	3.9	35
3336	Inherited GINS1 deficiency underlies growth retardation along with neutropenia and NK cell deficiency. Journal of Clinical Investigation, 2017, 127, 1991-2006.	3.9	115
3337	Inborn errors in RNA polymerase III underlie severe varicella zoster virus infections. Journal of Clinical Investigation, 2017, 127, 3543-3556.	3.9	125
3338	Evolutionary history of metastatic breast cancer reveals minimal seeding from axillary lymph nodes. Journal of Clinical Investigation, 2018, 128, 1355-1370.	3.9	123
3339	VarGenius executes cohort-level DNA-seq variant calling and annotation and allows to manage the resulting data through a PostgreSQL database. BMC Bioinformatics, 2018, 19, 477.	1.2	23
3340	Computational identification of deleterious synonymous variants in human genomes using a feature-based approach. BMC Medical Genomics, 2019, 12, 12.	0.7	47
3341	Case Report: Novel mutations in TBC1D24 are associated with autosomal dominant tonic-clonic and myoclonic epilepsy and recessive Parkinsonism, psychosis, and intellectual disability. F1000Research, 2017, 6, 553.	0.8	24
3342	Recent advances in predicting geneâ€“disease associations. F1000Research, 2017, 6, 578.	0.8	36
3343	Clinotator: analyzing ClinVar variation reports to prioritize reclassification efforts. F1000Research, 2018, 7, 462.	0.8	3
3344	Clinotator: analyzing ClinVar variation reports to prioritize reclassification efforts. F1000Research, 2018, 7, 462.	0.8	2
3345	Rare variants of the 3â€™-5â€™ DNA exonuclease TREX1 in early onset small vessel stroke. Wellcome Open Research, 2017, 2, 106.	0.9	7
3346	Erosion of Conserved Binding Sites in Personal Genomes Points to Medical Histories. PLoS Computational Biology, 2016, 12, e1004711.	1.5	7
3347	PredictSNP2: A Unified Platform for Accurately Evaluating SNP Effects by Exploiting the Different Characteristics of Variants in Distinct Genomic Regions. PLoS Computational Biology, 2016, 12, e1004962.	1.5	149

#	Article	IF	Citations
3348	Predicted Molecular Effects of Sequence Variants Link to System Level of Disease. PLoS Computational Biology, 2016, 12, e1005047.	1.5	14
3349	Multiscale mutation clustering algorithm identifies pan-cancer mutational clusters associated with pathway-level changes in gene expression. PLoS Computational Biology, 2017, 13, e1005347.	1.5	14
3350	Mendel,MD: A user-friendly open-source web tool for analyzing WES and WGS in the diagnosis of patients with Mendelian disorders. PLoS Computational Biology, 2017, 13, e1005520.	1.5	10
3351	High-confidence assessment of functional impact of human mitochondrial non-synonymous genome variations by APOGEE. PLoS Computational Biology, 2017, 13, e1005628.	1.5	54
3352	Exome Sequencing of Phenotypic Extremes Identifies CAV2 and TMC6 as Interacting Modifiers of Chronic Pseudomonas aeruginosa Infection in Cystic Fibrosis. PLoS Genetics, 2015, 11, e1005273.	1.5	39
3353	Heterogeneity of Human Neutrophil CD177 Expression Results from CD177P1 Pseudogene Conversion. PLoS Genetics, 2016, 12, e1006067.	1.5	36
3354	Rare Functional Variant in TM2D3 is Associated with Late-Onset Alzheimer's Disease. PLoS Genetics, 2016, 12, e1006327.	1.5	47
3355	Somatic Mutation Patterns in Hemizygous Genomic Regions Unveil Purifying Selection during Tumor Evolution. PLoS Genetics, 2016, 12, e1006506.	1.5	24
3356	Predicting the impact of Lynch syndrome-causing missense mutations from structural calculations. PLoS Genetics, 2017, 13, e1006739.	1.5	90
3357	Genome-wide association study of red blood cell traits in Hispanics/Latinos: The Hispanic Community Health Study/Study of Latinos. PLoS Genetics, 2017, 13, e1006760.	1.5	53
3358	Identifying and exploiting trait-relevant tissues with multiple functional annotations in genome-wide association studies. PLoS Genetics, 2018, 14, e1007186.	1.5	30
3359	Exome-wide somatic mutation characterization of small bowel adenocarcinoma. PLoS Genetics, 2018, 14, e1007200.	1.5	62
3360	PEA15 loss of function and defective cerebral development in the domestic cat. PLoS Genetics, 2020, 16, e1008671.	1.5	4
3361	A complementary study approach unravels novel players in the pathoetiology of Hirschsprung disease. PLoS Genetics, 2020, 16, e1009106.	1.5	7
3362	Nationwide germline whole genome sequencing of 198 consecutive pediatric cancer patients reveals a high incidence of cancer prone syndromes. PLoS Genetics, 2020, 16, e1009231.	1.5	64
3363	The Subclonal Architecture of Metastatic Breast Cancer: Results from a Prospective Community-Based Rapid Autopsy Program â€œCASCADEâ€• PLoS Medicine, 2016, 13, e1002204.	3.9	119
3364	Genome Sequencing of Idiopathic Pulmonary Fibrosis in Conjunction with a Medical School Human Anatomy Course. PLoS ONE, 2014, 9, e106744.	1.1	9
3365	PON-P2: Prediction Method for Fast and Reliable Identification of Harmful Variants. PLoS ONE, 2015, 10, e0117380.	1.1	180

#	Article	IF	Citations
3366	In-Silico Analysis of Inflammatory Bowel Disease (IBD) GWAS Loci to Novel Connections. PLoS ONE, 2015, 10, e0119420.	1.1	23
3367	ExScalibur: A High-Performance Cloud-Enabled Suite for Whole Exome Germline and Somatic Mutation Identification. PLoS ONE, 2015, 10, e0135800.	1.1	12
3368	ENTPRISE: An Algorithm for Predicting Human Disease-Associated Amino Acid Substitutions from Sequence Entropy and Predicted Protein Structures. PLoS ONE, 2016, 11, e0150965.	1.1	23
3369	Genetic Candidate Variants in Two Multigenerational Families with Childhood Apraxia of Speech. PLoS ONE, 2016, 11, e0153864.	1.1	33
3370	Incorporating Non-Coding Annotations into Rare Variant Analysis. PLoS ONE, 2016, 11, e0154181.	1.1	10
3371	Genetic Analysis of PLA2G6 in 22 Indian Families with Infantile Neuroaxonal Dystrophy, Atypical Late-Onset Neuroaxonal Dystrophy and Dystonia Parkinsonism Complex. PLoS ONE, 2016, 11, e0155605.	1.1	26
3372	Next-Generation Sequencing and In Vitro Expression Study of ADAMTS13 Single Nucleotide Variants in Deep Vein Thrombosis. PLoS ONE, 2016, 11, e0165665.	1.1	7
3373	Novel Candidate Genes and a Wide Spectrum of Structural and Point Mutations Responsible for Inherited Retinal Dystrophies Revealed by Exome Sequencing. PLoS ONE, 2016, 11, e0168966.	1.1	40
3374	Rare Synaptogenesis-Impairing Mutations in SLITRK5 Are Associated with Obsessive Compulsive Disorder. PLoS ONE, 2017, 12, e0169994.	1.1	25
3375	Expression Quantitative Trait loci (QTL) in tumor adjacent normal breast tissue and breast tumor tissue. PLoS ONE, 2017, 12, e0170181.	1.1	12
3376	Molecular modeling and molecular dynamic simulation of the effects of variants in the TGFBR2 kinase domain as a paradigm for interpretation of variants obtained by next generation sequencing. PLoS ONE, 2017, 12, e0170822.	1.1	19
3377	Exome-wide association study reveals novel susceptibility genes to sporadic dilated cardiomyopathy. PLoS ONE, 2017, 12, e0172995.	1.1	92
3378	Whole-genome sequencing suggests mechanisms for 22q11.2 deletion-associated Parkinsonâ€™s disease. PLoS ONE, 2017, 12, e0173944.	1.1	17
3379	Epigenomic annotation of noncoding mutations identifies mutated pathways in primary liver cancer. PLoS ONE, 2017, 12, e0174032.	1.1	9
3380	Multiple components of PKA and TGF-Î² pathways are mutated in pseudomyxoma peritonei. PLoS ONE, 2017, 12, e0174898.	1.1	15
3381	Rare variants of small effect size in neuronal excitability genes influence clinical outcome in Japanese cases of SCN1A truncation-positive Dravet syndrome. PLoS ONE, 2017, 12, e0180485.	1.1	18
3382	Novel and ultra-rare damaging variants in neuropeptide signaling are associated with disordered eating behaviors. PLoS ONE, 2017, 12, e0181556.	1.1	37
3383	Whole gene sequencing identifies deep-intronic variants with potential functional impact in patients with hypertrophic cardiomyopathy. PLoS ONE, 2017, 12, e0182946.	1.1	41

#	Article	IF	Citations
3384	Telomere biology and telomerase mutations in cirrhotic patients with hepatocellular carcinoma. PLoS ONE, 2017, 12, e0183287.	1.1	20
3385	Mutational analysis of ITPR1 in a Taiwanese cohort with cerebellar ataxias. PLoS ONE, 2017, 12, e0187503.	1.1	13
3386	CELSR2 is a candidate susceptibility gene in idiopathic scoliosis. PLoS ONE, 2017, 12, e0189591.	1.1	17
3387	Airway ciliary dysfunction and respiratory symptoms in patients with transposition of the great arteries. PLoS ONE, 2018, 13, e0191605.	1.1	17
3388	Generalising better: Applying deep learning to integrate deleteriousness prediction scores for whole-exome SNV studies. PLoS ONE, 2018, 13, e0192829.	1.1	14
3389	Identifying genetic variants associated with ritodrine-induced pulmonary edema. PLoS ONE, 2020, 15, e0241215.	1.1	4
3390	Genome-wide linkage analysis of families with primary hyperhidrosis. PLoS ONE, 2020, 15, e0244565.	1.1	7
3391	CARD9-Dependent Neutrophil Recruitment Protects against Fungal Invasion of the Central Nervous System. PLoS Pathogens, 2015, 11, e1005293.	2.1	184
3392	Whole genome sequencing of extreme phenotypes identifies variants in CD101 and UBE2V1 associated with increased risk of sexually acquired HIV-1. PLoS Pathogens, 2017, 13, e1006703.	2.1	16
3393	PSEN1 p.Met233Val in a Complex Neurodegenerative Movement and Neuropsychiatric Disorder. Journal of Movement Disorders, 2018, 11, 45-48.	0.7	12
3394	Glycogen storage disease type VI can progress to cirrhosis: ten Chinese patients with GSD VI and a literature review. Journal of Pediatric Endocrinology and Metabolism, 2020, 33, 1321-1333.	0.4	7
3395	A comprehensive in silico analysis, distribution and frequency of human Nkx2-5 mutations; A critical gene in congenital heart disease. Journal of Cardiovascular and Thoracic Research, 2019, 11, 287-299.	0.3	8
3397	Genetic polymorphisms and transcription profiles associated with intracranial aneurysm: a key role for NOTCH3. Aging, 2019, 11, 5173-5191.	1.4	3
3398	Clonal and microclonal mutational heterogeneity in high hyperdiploid acute lymphoblastic leukemia. Oncotarget, 2016, 7, 72733-72745.	0.8	12
3399	Charcot-Marie-Tooth gene, SBF2, associated with taxane-induced peripheral neuropathy in African Americans. Oncotarget, 2016, 7, 82244-82253.	0.8	35
3400	Whole-exome and transcriptome sequencing of refractory diffuse large B-cell lymphoma. Oncotarget, 2016, 7, 86433-86445.	0.8	43
3401	Characterization of RNA editome in primary and metastatic lung adenocarcinomas. Oncotarget, 2017, 8, 11517-11529.	0.8	7
3402	HIPK2-T566 autophosphorylation diversely contributes to UV- and doxorubicin-induced HIPK2 activation. Oncotarget, 2017, 8, 16744-16754.	0.8	6

#	Article	IF	Citations
3403	A comprehensive genomic meta-analysis identifies confirmatory role ofOBSCNgene in breast tumorigenesis. Oncotarget, 2017, 8, 102263-102276.	0.8	23
3404	Fanconi anemia and homologous recombination gene variants are associated with functional DNA repair defects in vitro and poor outcome in patients with advanced head and neck squamous cell carcinoma. Oncotarget, 2018, 9, 18198-18213.	0.8	37
3405	The search for cis-regulatory driver mutations in cancer genomes. Oncotarget, 2015, 6, 32509-32525.	0.8	18
3406	Germline genetics of cancer of unknown primary (CUP) and its specific subtypes. Oncotarget, 2016, 7, 22140-22149.	0.8	12
3407	Mutation based treatment recommendations from next generation sequencing data: a comparison of web tools. Oncotarget, 2016, 7, 22064-22076.	0.8	10
3408	The role of genomics and genetics in pulmonary arterial hypertension. Global Cardiology Science & Practice, 2020, 2020, e202013.	0.3	5
3409	Noncoding Variants Functional Prioritization Methods Based on Predicted Regulatory Factor Binding Sites. Current Genomics, 2017, 18, 322-331.	0.7	4
3410	Whole Exome Sequencing Reveals a BSCL2 Mutation Causing Progressive Encephalopathy with Lipodystrophy (PELD) in an Iranian Pediatric Patient. Iranian Biomedical Journal, 2016, 20, 295-301.	0.4	8
3411	Whole Exome Sequencing Reveals a XPNPEP3 Novel Mutation Causing Nephronophthisis in a Pediatric Patient. Iranian Biomedical Journal, 2020, 24, 400-403.	0.4	4
3412	Novel LMNA mutations in Greek and Myanmar Patients with Progeroid Features and Cardiac Manifestations. Aging Pathobiology and Therapeutics, 2020, 2, 101-105.	0.3	3
3413	A Bibliometric Insight of Genetic Factors in ASD: Emerging Trends and New Developments. Brain Sciences, 2021, 11, 33.	1.1	11
3414	Pharmacogenomics, How to Deal with Different Types of Variants in Next Generation Sequencing Data in the Personalized Medicine Area. Journal of Clinical Medicine, 2021, 10, 34.	1.0	4
3415	Increased burden of rare deleterious variants of the KCNQ1 gene in patients with largeâ€'vessel ischemic stroke. Molecular Medicine Reports, 2019, 19, 3263-3272.	1.1	3
3416	Genetic interactions between ANLN and KDR are prognostic for breast cancer survival. Oncology Reports, 2019, 42, 2255-2266.	1.2	16
3417	APEX1 Polymorphism and Mercaptopurine-Related Early Onset Neutropenia in Pediatric Acute Lymphoblastic Leukemia. Cancer Research and Treatment, 2018, 50, 823-834.	1.3	14
3418	Population Prevalence of Deleterious <i>SGCE</i> Variants. Tremor and Other Hyperkinetic Movements, 2020, 10, 50.	1.1	4
3419	Molecular genetic decoding of malformations of cortical development. Journal of Genetic Medicine, 2015, 12, 12-18.	0.1	2
3420	A network of epigenetic modifiers and DNA repair genes controls tissue-specific copy number alteration preference. ELife, 2016, 5, .	2.8	8

#	Article	IF	Citations
3421	Moyamoya disease factor RNF213 is a giant E3 ligase with a dynein-like core and a distinct ubiquitin-transfer mechanism. ELife, 2020, 9, .	2.8	74
3422	Patient-specific genomics and cross-species functional analysis implicate LRP2 in hypoplastic left heart syndrome. ELife, 2020, 9, .	2.8	29
3423	Systematic identification of cis-regulatory variants that cause gene expression differences in a yeast cross. ELife, 2020, 9, .	2.8	18
3424	ShapeGTB: the role of local DNA shape in prioritization of functional variants in human promoters with machine learning. PeerJ, 2018, 6, e5742.	0.9	2
3425	PLA2G6 Mutations Related to Distinct Phenotypes: A New Case with Early-onset Parkinsonism. Tremor and Other Hyperkinetic Movements, 2016, 6, 363.	1.1	11
3426	Machine learning meets omics: applications and perspectives. Briefings in Bioinformatics, 2022, 23, .	3.2	50
3427	Whole Exome Sequencing in Patients with Phenotypically Associated Familial Intracranial Aneurysm. Korean Journal of Radiology, 2022, 23, 101.	1.5	7
3428	In Silico Analysis of Novel Titin Non-Synonymous Missense Variants Detected by Targeted Next-Generation Sequencing in a Cohort of Romanian Index Patients with Hypertrophic Cardiomyopathy. Revista Romana De Cardiologie, 2021, 31, 565-571.	0.0	1
3429	Rare germline variants in childhood cancer patients suspected of genetic predisposition to cancer. Genes Chromosomes and Cancer, 2022, 61, 81-93.	1.5	2
3430	Biallelic FRA10AC1 variants cause a neurodevelopmental disorder with growth retardation. Brain, 2022, 145, 1551-1563.	3.7	9
3431	KCTD7-related progressive myoclonic epilepsy: report of three Indian families and review of literature. Clinical Dysmorphology, 2022, 31, 6-10.	0.1	4
3432	A Novel Variant of <scp>ATP5MC3</scp> Associated with Both Dystonia and Spastic Paraplegia. Movement Disorders, 2022, 37, 375-383.	2.2	10
3434	Absence of the RNF213 p.R4810K variant may indicate a severe form of pediatric moyamoya disease in Japanese patients. Journal of Neurosurgery: Pediatrics, 2022, 29, 48-56.	0.8	10
3435	Novel alterations in IFT172 and KIFAP3 may induce basal cell carcinoma. Orphanet Journal of Rare Diseases, 2021, 16, 443.	1.2	2
3436	The genetic impact of an Ebola outbreak on a wild gorilla population. BMC Genomics, 2021, 22, 735.	1.2	2
3437	Comparison of in silico strategies to prioritize rare genomic variants impacting RNA splicing for the diagnosis of genomic disorders. Scientific Reports, 2021, 11, 20607.	1.6	37
3438	Deciphering the genetic architecture and ethnographic distribution of IRD in three ethnic populations by whole genome sequence analysis. PLoS Genetics, 2021, 17, e1009848.	1.5	13
3439	Segmentation and genome annotation algorithms for identifying chromatin state and other genomic patterns. PLoS Computational Biology, 2021, 17, e1009423.	1.5	21

#	Article	IF	Citations
3440	Clinical evolution, genetic landscape and trajectories of clonal hematopoiesis in SAMD9/SAMD9L syndromes. Nature Medicine, 2021, 27, 1806-1817.	15.2	79
3441	AI applications in functional genomics. Computational and Structural Biotechnology Journal, 2021, 19, 5762-5790.	1.9	34
3442	Combinatorial patterns of gene expression changes contribute to variable expressivity of the developmental delay-associated 16p12.1 deletion. Genome Medicine, 2021, 13, 163.	3.6	5
3443	Clinical and genetic characterization of adultâ€onset leukoencephalopathy caused by CSF1R mutations. Annals of Clinical and Translational Neurology, 2021, 8, 2121-2131.	1.7	9
3445	Comprehensive multi-omics integration identifies differentially active enhancers during human brain development with clinical relevance. Genome Medicine, 2021, 13, 162.	3.6	9
3446	Which Is the Best In Silico Program for the Missense Variations in IDUA Gene? A Comparison of 33 Programs Plus a Conservation Score and Evaluation of 586 Missense Variants. Frontiers in Molecular Biosciences, 2021, 8, 752797.	1.6	5
3447	Prevalence and Clinical Characteristics of Hearing Loss Caused by MYH14 Variants. Genes, 2021, 12, 1623.	1.0	5
3448	T and B cell abnormalities, pneumocystis pneumonia, and chronic lymphocytic leukemia associated with an AIOLOS defect in patients. Journal of Experimental Medicine, 2021, 218, .	4.2	18
3449	Disruptive NADSYN1 Variants Implicated in Congenital Vertebral Malformations. Genes, 2021, 12, 1615.	1.0	7
3450	Role of ADAMTS13, VWF and F8 genes in deep vein thrombosis. PLoS ONE, 2021, 16, e0258675.	1.1	6
3451	Structural and functional analysis of somatic coding and UTR indels in breast and lung cancer genomes. Scientific Reports, 2021, 11, 21178.	1.6	1
3452	Network-Based Analysis to Identify Drivers of Metastatic Prostate Cancer Using GoNetic. Cancers, 2021, 13, 5291.	1.7	2
3453	Clinical manifestations of patients with <scp>GDF2</scp> mutations associated with hereditary hemorrhagic telangiectasia type 5. American Journal of Medical Genetics, Part A, 2022, 188, 199-209.	0.7	7
3454	Leveraging auxiliary data from arbitrary distributions to boost GWAS discovery with Flexible cFDR. PLoS Genetics, 2021, 17, e1009853.	1.5	3
3455	Identification of Novel and Recurrent Variants in MYO15A in Ashkenazi Jewish Patients With Autosomal Recessive Nonsyndromic Hearing Loss. Frontiers in Genetics, 2021, 12, 737782.	1.1	1
3457	In-vitro characterization of coding variants with predicted functional implications in the efflux transporter multidrug resistance protein 4 (MRP4, ABCC4). Pharmacogenetics and Genomics, 2021, Publish Ahead of Print, .	0.7	2
3460	Potential of different cells-derived exosomal microRNA cargos for treating spinal cord injury. Journal of Orthopaedic Translation, 2021, 31, 33-40.	1.9	14
3461	Standardisation of pathogenicity classification for somatic alterations in solid tumours and haematologic malignancies. European Journal of Cancer, 2021, 159, 1-15.	1.3	7

#	Article	IF	Citations
3465	MICO: A meta-tool for prediction of the effects of non-synonymous mutations. Bioinformation, 2014, 10, 469-471.	0.2	0
3478	Computational approaches for identifying deleterious nonsynonymous variants from human genomes. , 2015, , .		1
3480	Translational Research Methods: Basics of Renal Molecular Biology. , 2016, , 425-445.		0
3483	Application of Genomics to the Study of Human Growth Disorders. Translational Bioinformatics, 2016, , 363-384.	0.0	0
3500	Whole exome and genome sequencing for Mendelian immune disorders: from molecular diagnostics to new disease variant and gene discovery. LymphoSign Journal, 0, , .	0.1	1
3501	Genetic and Genomic Approaches to Acute Lung Injury. Respiratory Medicine, 2017, , 133-159.	0.1	0
3503	Application of a next-generation genetic analysis in the field of hemophilia. Japanese Journal of Thrombosis and Hemostasis, 2017, 28, 24-32.	0.1	0
3504	Genomic Analysis and In Vivo Functional Validation of Brain Somatic Mutations Leading to Focal Cortical Malformations. Neuromethods, 2017, , 299-327.	0.2	0
3522	Genetic analysis of single-minded 1 gene in early-onset severely obese children and adolescents. PLoS ONE, 2017, 12, e0177222.	1.1	4
3546	Whole-exome sequencing for detecting inborn errors of immunity: overview and perspectives. F1000Research, 2017, 6, 2056.	0.8	3
3550	Germline Genetic IKZF1 Variation and Predisposition to Childhood Acute Lymphoblastic Leukemia. SSRN Electronic Journal, 0, , .	0.4	0
3551	A Multiplexed Assay for Exon Recognition Reveals That an Unappreciated Fraction of Rare Genetic Variants Cause Large-Effect Disruptions to Splicing. SSRN Electronic Journal, 0, , .	0.4	0
3553	Mutations in the Glycosyltransferase Domain of GLT8D1 Cause Amyotrophic Lateral Sclerosis. SSRN Electronic Journal, 0, , .	0.4	0
3554	Computational Prediction of Driver Missense Mutations in Melanoma. Lecture Notes in Computer Science, 2018, , 438-447.	1.0	0
3556	Undifferentiated Sarcomas Develop Through Distinct Evolutionary Pathways. SSRN Electronic Journal, 0, , .	0.4	0
3564	SEARCHING FOR MISSING HERITABILITY FOR BREAST CANCER BY WHOLE EXOME SEQUENCING: EVALUATION OF VARIANT PATHOGENICITY AND SELECTION OF CANDIDATE MUTATIONS. Voprosy Onkologii, 2018, 64, 310-318.	0.1	0
3581	Whole exome analysis of primary immunodeficiency. Vavilovskii Zhurnal Genetiki I Selektsii, 2018, 22, 620-626.	0.4	0
3606	Geleophysic dysplasia: one gene, different phenotypes. Ukrainian Journal of Pediatric Endocrinology, 2018, .	0.1	0

#	Article	IF	Citations
3611	Genomic Revolution-Driven Cancer Research. , 2019, , 39-60.		0
3612	Guidelines for Bioinformatics and the Statistical Analysis of Omic Data. , 2019, , 45-75.		0
3616	Megalencephalic Leukoencephalopathy with Subcortical Cysts: Presentation of an Asymptomatic Patient and Review of Literature. Iranian Journal of Pediatrics, 2019, In Press, .	0.1	0
3663	CERENKOV3: Clustering and molecular network-derived features improve computational prediction of functional noncoding SNPs. , 2019, , .		0
3667	Hadoop and PySpark for reproducibility and scalability of genomic sequencing studies. , 2019, , .		0
3676	Combining eQTL and SNP Annotation Data to Identify Functional Noncoding SNPs in GWAS Trait-Associated Regions. Methods in Molecular Biology, 2020, 2082, 73-86.	0.4	2
3685	Exomes of Ductal Luminal Breast Cancer Patients from Southwest Colombia: Gene Mutational Profile and Related Expression Alterations. Biomolecules, 2020, 10, 698.	1.8	4
3688	Once in a Blue Moon, a Very Rare Coexistence of Glutaric Acidemia Type I and Mucopolysaccharidosis Type IIIB in a Patient. Iranian Biomedical Journal, 2020, 24, 201-205.	0.4	2
3689	Germline Mutation in MUS81 Resulting in Impaired Protein Stability is Associated with Familial Breast and Thyroid Cancer. Cancers, 2020, 12, 1289.	1.7	3
3693	Bioinformatics in Clinical Genomic Sequencing. Clinics in Laboratory Medicine, 2020, 40, 163-187.	0.7	5
3698	Characterization of a novel mutation in the MYOC gene in a Chinese family with primary openâ€‘angle glaucoma. Molecular Medicine Reports, 2020, 22, 3263-3270.	1.1	3
3704	Detection of PKD1 and PKD2 Somatic Variants in Autosomal Dominant Polycystic Kidney Cyst Epithelial Cells by Whole-Genome Sequencing. Journal of the American Society of Nephrology: JASN, 2021, 32, 3114-3129.	3.0	13
3705	Germline POT1 Deregulation Can Predispose to Myeloid Malignancies in Childhood. International Journal of Molecular Sciences, 2021, 22, 11572.	1.8	8
3706	A novel synonymous ABCA3 variant identified in a Chinese family with lethal neonatal respiratory failure. BMC Medical Genomics, 2021, 14, 256.	0.7	2
3707	Genome-Wide Landscape of North-Eastern European Populations: A View from Lithuania. Genes, 2021, 12, 1730.	1.0	3
3708	Whole-Exome Sequencing in Papillary Microcarcinoma: Potential Early Biomarkers of Lateral Lymph Node Metastasis. Endocrinology and Metabolism, 2021, 36, 1086-1094.	1.3	4
3709	<scp>PUS3</scp>â€related disorder: Report of a novel patient and delineation of the phenotypic spectrum. American Journal of Medical Genetics, Part A, 2022, 188, 635-641.	0.7	5
3710	A missense mutation converts the Na+,K+-ATPase into an ion channel and causes therapy-resistant epilepsy. Journal of Biological Chemistry, 2021, 297, 101355.	1.6	9

#	Article	IF	Citations
3711	Whole-exome analysis in Tunisian Imazighen and Arabs shows the impact of demography in functional variation. Scientific Reports, 2021, 11, 21125.	1.6	4
3712	Neonatal Manifestations of Chronic Granulomatous Disease: MAS/HLH and Necrotizing Pneumonia as Unusual Phenotypes and Review of the Literature. Journal of Clinical Immunology, 2022, 42, 299-311.	2.0	11
3717	Marfan syndrome: whole-exome sequencing reveals de novo mutations, second gene and genotypeâ€“phenotype correlations in the Chinese population. Bioscience Reports, 2020, 40, .	1.1	8
3722	Vertical transmission of a large calvarial ossification defect due to heterozygous variants of ALX4 and TWIST1. American Journal of Medical Genetics, Part A, 2021, 185, 916-922.	0.7	1
3723	Interpretation of Biological Data at a Glance. Journal of Biotechnology and Bioindustry, 2020, 8, 38-47.	0.1	0
3724	Phenotypic differences of mutationâ€negative cases in Gitelman syndrome clinically diagnosed in adulthood. Human Mutation, 2021, 42, 300-309.	1.1	4
3726	Calibrating variant-scoring methods for clinical decision making. Bioinformatics, 2021, 36, 5709-5711.	1.8	10
3728	ncVarDB: a manually curated database for pathogenic non-coding variants and benign controls. Database: the Journal of Biological Databases and Curation, 2020, 2020, .	1.4	8
3730	Functional Evolutionary Modeling Exposes Overlooked Protein-Coding Genes Involved in Cancer. Lecture Notes in Computer Science, 2020, , 119-126.	1.0	0
3749	M-DATA: A statistical approach to jointly analyzing de novo mutations for multiple traits. PLoS Genetics, 2021, 17, e1009849.	1.5	4
3750	Network analysis reveals rare disease signatures across multiple levels of biological organization. Nature Communications, 2021, 12, 6306.	5.8	36
3751	A functional variant of SHARPIN confers increased risk of late-onset Alzheimerâ€™s disease. Journal of Human Genetics, 2022, 67, 203-208.	1.1	8
3752	Identification of Functional Genetic Determinants of Cardiac Troponin T and I in a Multiethnic Population and Causal Associations With Atrial Fibrillation. Circulation Genomic and Precision Medicine, 2021, 14, CIRCGEN121003460.	1.6	5
3757	A Hidden Structural Variation in a Known IRD Gene: A Cautionary Tale of Two New Disease Candidate Genes. Journal of Physical Education and Sports Management, 2021, , mcs.a006131.	0.5	0
3759	Myoclonic status epilepticus and cerebellar hypoplasia associated with a novel variant in the GRIA3 gene. Neurogenetics, 2021, , 1.	0.7	2
3760	VPMBench: a test bench for variant prioritization methods. BMC Bioinformatics, 2021, 22, 543.	1.2	0
3763	Optimization of Biogas Production in a Batch Laboratory Digester Using Total Solids, Substrate Retention Time, and Mesophilic Temperature. International Journal of Power and Energy Research, 2020, 4, .	0.4	0
3771	A weighted empirical Bayes risk prediction model using multiple traits. Statistical Applications in Genetics and Molecular Biology, 2020, 19, .	0.2	0

#	Article	IF	Citations
3772	Duchenne Muscular Dystrophy and Early Onset Hypertrophic Cardiomyopathy associated with Mutations in Dystrophin and Hypertrophic Cardiomyopathy-Associated Genes. Journal of Pediatric Genetics, 0, , .	0.3	0
3773	Kidney Stones Are Prevalent in Individuals with Pseudoxanthoma Elasticum, a Genetic Ectopic Mineralization Disorder. International Journal of Dermatology and Venereology, 2020, 3, 198-204.	0.1	5
3779	Digenic Variants in the FGF21 Signaling Pathway Associated with Severe Insulin Resistance and Pseudoacromegaly. Journal of the Endocrine Society, 2020, 4, bvaa138.	0.1	6
3781	Identification of Novel Functional Variants of SIN3A and SRSF1 among Somatic Variants in Acute Myeloid Leukemia Patients. Molecules and Cells, 2018, 41, 465-475.	1.0	4
3782	Autosomal Recessive Nonsyndromic Hearing Loss: A Case Report with a Mutation in TRIOBP Gene. International Journal of Molecular and Cellular Medicine, 2015, 4, 245-7.	1.1	5
3783	Mutation spectrum of the Norrie disease pseudoglioma (NDP) gene in Indian patients with FEVR. Molecular Vision, 2016, 22, 491-502.	1.1	14
3784	Functional Analysis of A Novel Splicing Mutation in The Mutase Gene of Two Unrelated Pedigrees. Cell Journal, 2016, 18, 397-404.	0.2	1
3785	Genetic Analysis of Iranian Patients with Familial Hypercholesterolemia. Iranian Biomedical Journal, 2018, 22, 117-22.	0.4	7
3786	Hereditary Myoclonus Dystonia: A Novel Variant and Phenotype Including Intellectual Disability. Tremor and Other Hyperkinetic Movements, 2018, 8, 547.	1.1	1
3787	Variants in the gene in a Brazilian population with Stargardt disease. Molecular Vision, 2018, 24, 546-559.	1.1	12
3788	Advanced molecular approaches pave the road to a clear-cut diagnosis of hereditary retinal dystrophies. Molecular Vision, 2018, 24, 679-689.	1.1	1
3789	Rare variant in gene causing congenital muscular dystrophy in a Sudanese family. A case report. Acta Myologica, 2019, 38, 21-24.	1.5	3
3790	CRB1-Related Leber Congenital Amaurosis: Reporting Novel Pathogenic Variants and a Brief Review on Mutations Spectrum. Iranian Biomedical Journal, 2019, 23, 362-8.	0.4	1
3791	Five major sequence variants and copy number variants in the gene account for one-third of Japanese patients with autosomal recessive and simplex retinitis pigmentosa. Molecular Vision, 2019, 25, 766-779.	1.1	7
3792	CERENKOV3: Clustering and molecular network-derived features improve computational prediction of functional noncoding SNPs. Pacific Symposium on Biocomputing Pacific Symposium on Biocomputing, 2020, 25, 535-546.	0.7	0
3793	Hadoop and PySpark for reproducibility and scalability of genomic sequencing studies. Pacific Symposium on Biocomputing Pacific Symposium on Biocomputing, 2020, 25, 523-534.	0.7	0
3794	Estrogen Receptors (ESRs) Mutations in Adolescent Idiopathic Scoliosis: A Cross-Sectional Study. Medical Science Monitor, 2020, 26, e921611.	0.5	2
3795	Noncoding mutation in contributes to inherited retinal degenerations. Molecular Vision, 2021, 27, 95-106.	1.1	2

#	Article	IF	Citations
3796	A novel homozygous missense mutation p.P388S in causes protein instability and retinitis pigmentosa. Molecular Vision, 2021, 27, 179-190.	1.1	3
3797	Performance of meta-predictors for the classification of MED13L missense variations, implication of raw parameters. European Journal of Medical Genetics, 2022, 65, 104398.	0.7	0
3798	Revisiting the neutral dynamics derived limiting guanine-cytosine content using human de novo point mutation data. Meta Gene, 2022, 31, 100994.	0.3	0
3799	Germline biallelic mutation affecting the transcription factor Helios causes pleiotropic defects of immunity. Science Immunology, 2021, 6, eabe3981.	5.6	16
3800	Whole-genome analysis of Nigerian patients with breast cancer reveals ethnic-driven somatic evolution and distinct genomic subtypes. Nature Communications, 2021, 12, 6946.	5.8	22
3801	The importance of automation in genetic diagnosis: Lessons from analyzing an inherited retinal degeneration cohort with the Mendelian Analysis Toolkit (MATK). Genetics in Medicine, 2022, 24, 332-343.	1.1	7
3802	Desmoplakin and periplakin genetically and functionally contribute to eosinophilic esophagitis. Nature Communications, 2021, 12, 6795.	5.8	23
3803	A selection pressure landscape for 870 human polygenic traits. Nature Human Behaviour, 2021, 5, 1731-1743.	6.2	17
3804	Quantifying prediction of pathogenicity for within-codon concordance (PM5) using 7541 functional classifications of BRCA1 and MSH2 missense variants. Genetics in Medicine, 2022, 24, 552-563.	1.1	5
3805	The Mutational Landscape of PTK7 in Congenital Scoliosis and Adolescent Idiopathic Scoliosis. Genes, 2021, 12, 1791.	1.0	5
3806	Glucocerebrosidase (GBA) gene variants in a multi-ethnic Asian cohort with Parkinsonâ€™s disease: mutational spectrum and clinical features. Journal of Neural Transmission, 2022, 129, 37-48.	1.4	18
3808	Expression level is a major modifier of the fitness landscape of a protein coding gene. Nature Ecology and Evolution, 2022, 6, 103-115.	3.4	24
3809	Loss-of-function mutation in IKZF2 leads to immunodeficiency with dysregulated germinal center reactions and reduction of MAIT cells. Science Immunology, 2021, 6, eabe3454.	5.6	30
3811	The Somatic Mutation Paradigm in Congenital Malformations: Hirschsprung Disease as a Model. International Journal of Molecular Sciences, 2021, 22, 12354.	1.8	3
3813	Nonâ€coding regulatory elements: Potential roles in disease and the case of epilepsy. Neuropathology and Applied Neurobiology, 2022, 48, .	1.8	14
3815	Insilico Functional Analysis of Genome-Wide Dataset From 17,000 Individuals Identifies Candidate Malaria Resistance Genes Enriched in Malaria Pathogenic Pathways. Frontiers in Genetics, 2021, 12, 676960.	1.1	1
3816	Whole genome sequencing and in vitro splice assays reveal genetic causes for inherited retinal diseases. Npj Genomic Medicine, 2021, 6, 97.	1.7	27
3818	Possible <scp>EIF2AK2</scp>â€Associated Stressâ€Related Neurological Decompensation with Combined Dystonia and Striatal Lesions. Movement Disorders Clinical Practice, 2022, 9, 240-244.	0.8	6

#	Article	IF	Citations
3819	Genomic Variation Prediction: A Summary From Different Views. Frontiers in Cell and Developmental Biology, 2021, 9, 795883.	1.8	3
3820	New developments in the field of genomic technologies and their relevance to conservation management. Conservation Genetics, 2022, 23, 217-242.	0.8	26
3821	Rare variant analysis in eczema identifies exonic variants in DUSP1, NOTCH4 and SLC9A4. Nature Communications, 2021, 12, 6618.	5.8	17
3822	Retroperitoneal paraganglioma with asymptomatic follicular lymphoma : a case report. Journal of the Endocrine Society, 2021, 5, bvab171.	0.1	1
3823	Variant profiling of colorectal adenomas from three patients of two families with MSH3-related adenomatous polyposis. PLoS ONE, 2021, 16, e0259185.	1.1	5
3824	Molecular analysis of ABCA4 gene in an Iranian cohort with Stargardt disease. Gene Reports, 2021, , 101450.	0.4	0
3825	Decoding the effects of synonymous variants. Nucleic Acids Research, 2021, 49, 12673-12691.	6.5	17
3826	The Greek Variant in APP Gene: The Phenotypic Spectrum of APP Mutations. International Journal of Molecular Sciences, 2021, 22, 12355.	1.8	1
3827	Pharmacogenomic analysis of a genetically distinct Indigenous population. Pharmacogenomics Journal, 2021, , .	0.9	4
3828	Whole genome sequencing in a Knobloch syndrome family confirms the molecular diagnosis. Ophthalmic Genetics, 2021, , 1-9.	0.5	0
3829	Excess Heritability Contribution of Alcohol Consumption Variants in the â€œMissing Heritabilityâ€•of Type 2 Diabetes Mellitus. International Journal of Molecular Sciences, 2021, 22, 12318.	1.8	0
3831	Identification of discriminative gene-level and protein-level features associated with pathogenic gain-of-function and loss-of-function variants. American Journal of Human Genetics, 2021, 108, 2301-2318.	2.6	21
3832	Germline RET Leu56Met Variant Is Likely Not Causative of Multiple Endocrine Neoplasia Type 2. Frontiers in Endocrinology, 2021, 12, 764512.	1.5	0
3833	Novel variants in the LRP4 underlying Cenani-Lenz Syndactyly syndrome. Journal of Human Genetics, 2021, , .	1.1	5
3834	Glycoprotein 2 in health and disease: lifting the veil. Genes and Environment, 2021, 43, 53.	0.9	3
3835	Combining Human Genetics of Multiple Sclerosis with Oxidative Stress Phenotype for Drug Repositioning. Pharmaceutics, 2021, 13, 2064.	2.0	3
3836	Novel pathogenic variants and quantitative phenotypic analyses of Robinow syndrome: WNT signaling perturbation and phenotypic variability. Human Genetics and Genomics Advances, 2022, 3, 100074.	1.0	14
3837	What makes a good prediction? Feature importance and beginning to open the black box of machine learning in genetics. Human Genetics, 2022, 141, 1515-1528.	1.8	18

#	Article	IF	Citations
3838	High Coverage Whole Genome Sequencing of the Expanded 1000 Genomes Project Cohort Including 602 Trios. SSRN Electronic Journal, 0, , .	0.4	10
3839	Computational Methods and Approaches in Pharmacogenomic Research. , 2022, , 53-83.		1
3840	Genome-Wide Association Study of Clinical Outcome After Aneurysmal Subarachnoid Haemorrhage: Protocol. Translational Stroke Research, 2022, 13, 565-576.	2.3	5
3841	Thirteen Independent Genetic Loci Associated with Preserved Processing Speed in a Study of Cognitive Resilience in 330,097 Individuals in the UK Biobank. Genes, 2022, 13, 122.	1.0	3
3842	Automated prediction of the clinical impact of structural copy number variations. Scientific Reports, 2022, 12, 555.	1.6	3
3843	Overcoming constraints on the detection of recessive selection in human genes from population frequency data. American Journal of Human Genetics, 2022, 109, 33-49.	2.6	5
3844	Genotype-phenotype correlation of Parkinson's disease with PRKN variants. Neurobiology of Aging, 2022, 114, 117-128.	1.5	13
3845	Aberrant interaction between mutated ADAMTSL2 and LTBP4 is associated with adolescent idiopathic scoliosis. Gene, 2022, 814, 146126.	1.0	5
3846	CS-GANomaly: A Supervised Anomaly Detection Approach with Ancillary Classifier GANs for Chromosome Images. , 2020, , .		3
3847	PPSNV: A Novel Predictor for Pathogenicity of Nonsynonymous SNV based on Ensemble Learning. , 2021, , .		0
3848	Recurrent de novo Single Point Mutation on the Gene Encoding Na ⁺/K ⁺ Pump Results in Epilepsy. SSRN Electronic Journal, 0, , .	0.4	0
3849	Integration of tumour sequencing and caseâ€“control data to assess pathogenicity of RAD51C missense variants in familial breast cancer. Npj Breast Cancer, 2022, 8, 10.	2.3	0
3850	De novo mutations identified by whole-genome sequencing implicate chromatin modifications in obsessive-compulsive disorder. Science Advances, 2022, 8, eabi6180.	4.7	5
3851	Genome-wide association study meta-analysis identifies three novel loci for circulating anti-MÃ¼llerian hormone levels in women. Human Reproduction, 2022, 37, 1069-1082.	0.4	13
3852	Altered gene expression levels of genes related to muscle function in adults with cerebral palsy. Tissue and Cell, 2022, 76, 101744.	1.0	2
3853	Whole-Exome Sequencing Implicates Neuronal Calcium Channel with Familial Atrial Fibrillation. Frontiers in Genetics, 2022, 13, 806429.	1.1	1
3854	Variants in Mitochondrial <scp>ATP</scp> Synthase Cause Variable Neurologic Phenotypes. Annals of Neurology, 2022, 91, 225-237.	2.8	12
3856	A somatic UBA2 variant preceded ETV6-RUNX1 in the concordant BCP-ALL of monozygotic twins. Blood Advances, 2022, 6, 2275-2289.	2.5	5

#	Article	IF	Citations
3857	Machine learning techniques for pathogenicity prediction of non-synonymous single nucleotide polymorphisms in human body. Journal of Ambient Intelligence and Humanized Computing, 0, , 1.	3.3	0
3858	Predicting and interpreting large-scale mutagenesis data using analyses of protein stability and conservation. Cell Reports, 2022, 38, 110207.	2.9	62
3859	A comparison on predicting functional impact of genomic variants. NAR Genomics and Bioinformatics, 2022, 4, lqab122.	1.5	12
3860	An update on clinical applications of iPSCs from a genomic point of view. , 2022, , 147-175.		0
3861	The Difference in the Proportions of Deleterious Variations within and between Populations Influences the Estimation of FST. Genes, 2022, 13, 194.	1.0	2
3862	Whole-Exome Sequencing Identifies a Novel Germline Variant in PTK7 Gene in Familial Colorectal Cancer. International Journal of Molecular Sciences, 2022, 23, 1295.	1.8	2
3863	Evaluation of phenotype-driven gene prioritization methods for Mendelian diseases. Briefings in Bioinformatics, 2022, 23, .	3.2	17
3864	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
3865	Germline Variants of CYBA and TRPM4 Predispose to Familial Colorectal Cancer. Cancers, 2022, 14, 670.	1.7	11
3866	Delineation of a novel neurodevelopmental syndrome associated with PAX5 haploinsufficiency. Human Mutation, 2022, 43, 461-470.	1.1	5
3867	Evaluating the relevance of sequence conservation in the prediction of pathogenic missense variants. Human Genetics, 2022, 141, 1649-1658.	1.8	3
3868	Haplotype-based inference of the distribution of fitness effects. Genetics, 2022, 220, .	1.2	1
3869	Whole-exome sequencing analysis of a Japanese patient with hyperinsulinemia and liver dysfunction. Journal of the Endocrine Society, 2022, 6, bvac008.	0.1	0
3870	Genetic determinants of telomere length from 109,122 ancestrally diverse whole-genome sequences in TOPMed. Cell Genomics, 2022, 2, 100084.	3.0	29
3871	OUP accepted manuscript. Briefings in Bioinformatics, 2022, 23, .	3.2	5
3872	Whole Genome Sequencing Unravels New Genetic Determinants of Early-Onset Familial Osteoporosis and Low BMD in Malta. Genes, 2022, 13, 204.	1.0	2
3873	Variant interpretation in molecular autopsy: a useful dilemma. International Journal of Legal Medicine, 2022, 136, 475-482.	1.2	9
3874	Genetic analysis of pancreatic phospholipase A2 (PLA2G1B) in patients with chronic pancreatitis. Pancreatology, 2022, 22, 244-247.	0.5	0

#	Article	IF	Citations
3875	Deconstructing a Syndrome: Genomic Insights Into PCOS Causal Mechanisms and Classification. Endocrine Reviews, 2022, 43, 927-965.	8.9	75
3877	Inherited human Apollo deficiency causes severe bone marrow failure and developmental defects. Blood, 2022, 139, 2427-2440.	0.6	14
3878	Whole-Genome and Transcriptome Sequencing Identified NOTCH2 and HES1 as Potential Markers of Response to Imatinib in Desmoid Tumor (Aggressive Fibromatosis): A Phase II Trial Study. Cancer Research and Treatment, 2022, 54, 1240-1255.	1.3	4
3879	Germline sequence variants contributing to cancer susceptibility in South African breast cancer patients of African ancestry. Scientific Reports, 2022, 12, 802.	1.6	4
3881	A case of juvenile-onset pheochromocytoma with <i>KIF1B</i> p.V1529M germline mutation. Endocrine Journal, 2022, 69, 705-716.	0.7	3
3882	Combining RSPH9 founder mutation screening and next-generation sequencing analysis is efficient for primary ciliary dyskinesia diagnosis in Saudi patients. Journal of Human Genetics, 2022, 67, 381-386.	1.1	5
3883	Impaired catabolism of free oligosaccharides due to MAN2C1 variants causes a neurodevelopmental disorder. American Journal of Human Genetics, 2022, 109, 345-360.	2.6	4
3885	Integrative Identification of Genetic Loci Jointly Influencing Diabetes-Related Traits and Sleep Traits of Insomnia, Sleep Duration, and Chronotypes. Biomedicines, 2022, 10, 368.	1.4	3
3886	Recessive variants in COL25A1 gene as novel cause of arthrogryposis multiplex congenita with ocular congenital cranial dysinnervation disorder. Human Mutation, 2022, 43, 487-498.	1.1	8
3887	Autosomal dominant sleep-related hypermotor epilepsy caused by a previously unreported CHRNA4 variant. European Journal of Medical Genetics, 2022, 65, 104444.	0.7	2
3888	The genetic architecture of language functional connectivity. NeuroImage, 2022, 249, 118795.	2.1	14
3889	Machine learning methods for prediction of cancer driver genes: a survey paper. Briefings in Bioinformatics, 2022, 23, .	3.2	15
3890	A dominant negative variant of RAB5B disrupts maturation of surfactant protein B and surfactant protein C. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	3.3	9
3891	Consistency of variant interpretations among bioinformaticians and clinical geneticists in hereditary cancer panels. European Journal of Human Genetics, 2022, 30, 378-383.	1.4	5
3892	CELSR1 Risk Alleles in Familial Bicuspid Aortic Valve and Hypoplastic Left Heart Syndrome. Circulation Genomic and Precision Medicine, 2022, 15, CIRCGEN121003523.	1.6	11
3893	Case Report: Two Families With HPDL Related Neurodegeneration. Frontiers in Genetics, 2022, 13, 780764.	1.1	4
3894	Genetic load: genomic estimates and applications in non-model animals. Nature Reviews Genetics, 2022, 23, 492-503.	7.7	82
3895	Analysis of missense variants in the human genome reveals widespread gene-specific clustering and improves prediction of pathogenicity. American Journal of Human Genetics, 2022, 109, 457-470.	2.6	29

#	Article	IF	Citations
3896	Boosting Schizophrenia Genetics by Utilizing Genetic Overlap With Brain Morphology. Biological Psychiatry, 2022, 92, 291-298.	0.7	20
3897	Broadening the spectrum of loss-of-function variants in NPR-C-related extreme tall stature. Journal of the Endocrine Society, 2022, 6, bvac019.	0.1	2
3898	Discovery of genomic loci of the human cerebral cortex using genetically informed brain atlases. Science, 2022, 375, 522-528.	6.0	31
3899	The Diverse Phenotype of Intestinal Dysmotility Secondary to ACTG2â€related Disorders. Journal of Pediatric Gastroenterology and Nutrition, 2022, 74, 575-581.	0.9	4
3900	Further delineation of familial polycystic ovary syndrome (PCOS) via <scp>wholeâ€exome</scp> sequencing: <scp>PCOS</scp>â€related rare <scp>FBN3</scp> and <scp>FN1</scp> gene variants are identified. Journal of Obstetrics and Gynaecology Research, 2022, 48, 1202-1211.	0.6	9
3901	DECIPHER: Supporting the interpretation and sharing of rare disease phenotypeâ€linked variant data to advance diagnosis and research. Human Mutation, 2022, , .	1.1	10
3902	Computational Assessment of the Expression-Modulating Potential for Non-Coding Variants. Genomics, Proteomics and Bioinformatics, 2023, 21, 662-673.	3.0	3
3903	De novo coding variants in the AGO1 gene cause a neurodevelopmental disorder with intellectual disability. Journal of Medical Genetics, 2022, 59, 965-975.	1.5	13
3904	Whole-Exome Sequencing Revealed a Pathogenic Nonsense Variant in theSLC19A2Gene in an Iranian Family with Thiamine-Responsive Megaloblastic Anemia. Laboratory Medicine, 2022, 53, 640-650.	0.8	0
3905	EPIMUTESTR: a nearest neighbor machine learning approach to predict cancer driver genes from the evolutionary action of coding variants. Nucleic Acids Research, 2022, 50, e70-e70.	6.5	7
3906	AllelePred: A Simple Allele Frequencies Ensemble Predictor for Different Single Nucleotide Variants. IEEE/ACM Transactions on Computational Biology and Bioinformatics, 2022, PP, 1-1.	1.9	0
3907	Novel RAB3GAP1 Mutation in the First Tunisian Family With Warburg Micro Syndrome. Journal		

#	Article	IF	Citations
3915	A framework to score the effects of structural variants in health and disease. Genome Research, 2022, 32, 766-777.	2.4	17
3916	PRUNE1 c.<scp>933G</scp>>A synonymous variant induces exon 7 skipping, disrupts the <scp>DHHA2</scp> domain, and leads to an atypical <scp>NMIHBA</scp> syndrome presentation: Case report and review of the literature. American Journal of Medical Genetics, Part A, 2022, 188, 1868-1874.	0.7	2
3917	The Molecular Tumor Board Portal supports clinical decisions and automated reporting for precision oncology. Nature Cancer, 2022, 3, 251-261.	5.7	44
3918	DVPred: a disease-specific prediction tool for variant pathogenicity classification for hearing loss. Human Genetics, 2022, 141, 401-411.	1.8	6
3919	Usher syndrome type IV: clinically and molecularly confirmed by novel ARSG variants. Human Genetics, 2022, 141, 1723-1738.	1.8	16
3920	The Role of Natural Polymorphic Variants of DNA Polymerase Î² in DNA Repair. International Journal of Molecular Sciences, 2022, 23, 2390.	1.8	8
3921	Mutational spectrum of syndromic genes in sporadic brain arteriovenous malformation. Chinese Neurosurgical Journal, 2022, 8, 4.	0.3	1
3922	Molecular profiling of melanocortin 4 receptor variants and agouti-related peptide interactions in morbid obese phenotype: a novel paradigm from molecular docking and dynamics simulations. Biologia (Poland), 2022, 77, 1481.	0.8	2
3923	Genetic associations with carotid intima-media thickness link to atherosclerosis with sex-specific effects in sub-Saharan Africans. Nature Communications, 2022, 13, 855.	5.8	10
3924	Integrative analysis prioritizes the relevant genes and risk factors for chronic venous disease. Journal of Vascular Surgery: Venous and Lymphatic Disorders, 2022, 10, 738-748.e5.	0.9	3
3925	Rheumatoid arthritis in a patient with compound heterozygous variants in the COL11A2 gene and progressive hearing loss. Medicine (United States), 2022, 101, e28828.	0.4	1
3927	Landscape of TP53 Alterations in Chronic Lymphocytic Leukemia via Data Mining Mutation Databases. Frontiers in Oncology, 2022, 12, 808886.	1.3	5
3928	PIGNencephalopathy: Characterizing the epileptology. Epilepsia, 2022, 63, 974-991.	2.6	4
3929	A machine learning approach based on ACMG/AMP guidelines for genomic variant classification and prioritization. Scientific Reports, 2022, 12, 2517.	1.6	26
3931	Phylogenetic inference of changes in amino acid propensities with single-position resolution. PLoS Computational Biology, 2022, 18, e1009878.	1.5	0
3932	SWAAT Bioinformatics Workflow for Protein Structure-Based Annotation of ADME Gene Variants. Journal of Personalized Medicine, 2022, 12, 263.	1.1	2
3933	Current and Future Approaches to Classify VUSs in LGMD-Related Genes. Genes, 2022, 13, 382.	1.0	3
3935	Machine-learning of complex evolutionary signals improves classification of SNVs. NAR Genomics and Bioinformatics, 2022, 4, lqac025.	1.5	4

#	Article	IF	Citations
3938	SOD-1 Variants in Amyotrophic Lateral Sclerosis: Systematic Re-Evaluation According to ACMG-AMP Guidelines. Genes, 2022, 13, 537.	1.0	7
3939	GENESIS: Gene-Specific Machine Learning Models for Variants of Uncertain Significance Found in Catecholaminergic Polymorphic Ventricular Tachycardia and Long QT Syndrome-Associated Genes. Circulation: Arrhythmia and Electrophysiology, 2022, 15, 101161CIRCEP121010326.	2.1	17
3940	Impact of deleterious missense PRKCI variants on structural and functional dynamics of protein. Scientific Reports, 2022, 12, 3781.	1.6	7
3941	TADAâ€"a machine learning tool for functional annotation-based prioritisation of pathogenic CNVs. Genome Biology, 2022, 23, 67.	3.8	4
3942	Synonymous mutation in adenosine triphosphatase copperâ€transporting beta causes enhanced exon skipping in Wilson disease. Hepatology Communications, 2022, 6, 1611-1619.	2.0	6
3943	Phenotypic and genotypic characterization of familial hypercholesterolemia in French adult and pediatric populations. Journal of Clinical Lipidology, 2022, 16, 298-305.	0.6	0
3944	Drosophila functional screening of de novo variants in autism uncovers damaging variants and facilitates discovery of rare neurodevelopmental diseases. Cell Reports, 2022, 38, 110517.	2.9	24
3945	A CTNNB1â€altered medulloblastoma shows the immunophenotypic, DNA methylation and transcriptomic profiles of SHHâ€activated, and not WNTâ€activated, medulloblastoma. Neuropathology and Applied Neurobiology, 2022, 48, e12815.	1.8	0
3947	Phenotypic and mutational spectrum of ROR2 â€related Robinow syndrome. Human Mutation, 2022, 43, 900-918.	1.1	8
3948	ABO O blood group as a risk factor for platelet reactivity in heparin-induced thrombocytopenia. Blood, 2022, 140, 274-284.	0.6	9
3950	Implementation of preemptive DNA sequenceâ€"based pharmacogenomics testing across a large academic medical center: The Mayo-Baylor RIGHT 10K Study. Genetics in Medicine, 2022, 24, 1062-1072.	1.1	28
3951	Non-Motor Symptoms in PLA2G6-Associated Dystonia-Parkinsonism: A Case Report and Literature Review. Journal of Clinical Medicine, 2022, 11, 1590.	1.0	1
3952	A genome-wide meta-analysis identifies 50 genetic loci associated with carpal tunnel syndrome. Nature Communications, 2022, 13, 1598.	5.8	8
3953	Update on CD164 and LMX1A genes to strengthen their causative role in autosomal dominant hearing loss. Human Genetics, 2022, 141, 445-453.	1.8	6
3954	CRISPR Detection and Research on Screening Mutant Gene TTN of Moyamoya Disease Family Based on Whole Exome Sequencing. Frontiers in Molecular Biosciences, 2022, 9, 846579.	1.6	2
3956	Whole genome sequences discriminate hereditary hemorrhagic telangiectasia phenotypes by non-HHT deleterious DNA variation. Blood Advances, 2022, 6, 3956-3969.	2.5	9
3957	Whole-genome sequencing reveals novel ethnicity-specific rare variants associated with Alzheimerâ€™s disease. Molecular Psychiatry, 2022, 27, 2554-2562.	4.1	14
3960	Genomic analysis of familial pancreatic cancers and intraductal papillary mucinous neoplasms: A crossâ€sectional study. Cancer Science, 2022, 113, 1821-1829.	1.7	5

#	Article	IF	Citations
3961	Integrating whole genome sequencing, methylation, gene expression, topologically associated domain information in regulatory mutation prediction: A study of follicular lymphoma. Computational and Structural Biotechnology Journal, 2022, 20, 1726-1742.	1.9	1
3962	Understanding signatures of positive natural selection in human zinc transporter genes. Scientific Reports, 2022, 12, 4320.	1.6	2
3963	The Genetic Diagnosis of Ultrarare DEEs: An Ongoing Challenge. Genes, 2022, 13, 500.	1.0	7
3964	Whole-genome analysis of human embryonic stem cells enables rational line selection based on genetic variation. Cell Stem Cell, 2022, 29, 472-486.e7.	5.2	27
3965	Comprehensive variant spectrum of the CNGA3 gene in patients affected by achromatopsia. Human Mutation, 2022, 43, 832-858.	1.1	8
3966	An expanded phenotype centric benchmark of variant prioritisation tools. Human Mutation, 2022, 43, 539-546.	1.1	9
3967	Novel dominant and recessive variants in human ROBO1 cause distinct neurodevelopmental defects through different mechanisms. Human Molecular Genetics, 2022, 31, 2751-2765.	1.4	3
3969	TransPPMP: predicting pathogenicity of frameshift and non-sense mutations by a Transformer based on protein features. Bioinformatics, 2022, 38, 2705-2711.	1.8	4
3971	Whole genome sequencing delineates regulatory, copy number, and cryptic splice variants in early onset cardiomyopathy. Npj Genomic Medicine, 2022, 7, 18.	1.7	14
3972	Inherited and De Novo Variation in Lithuanian Genomes: Introduction to the Analysis of the Generational Shift. Genes, 2022, 13, 569.	1.0	3
3973	A multi-dimensional integrative scoring framework for predicting functional variants in the human genome. American Journal of Human Genetics, 2022, 109, 446-456.	2.6	18
3974	Signatures of genetic variation in human microRNAs point to processes of positive selection and population-specific disease risks. Human Genetics, 2022, 141, 1673-1693.	1.8	3
3976	Rare missense mutations in ABCA7 might increase Alzheimerâ€™s disease risk by plasma membrane exclusion. Acta Neuropathologica Communications, 2022, 10, 43.	2.4	11
3977	A neurodegenerative disease landscape of rare mutations in Colombia due to founder effects. Genome Medicine, 2022, 14, 27.	3.6	16
3978	The contribution of evolutionarily volatile promoters to molecular phenotypes and human trait variation. Genome Biology, 2022, 23, 89.	3.8	4
3979	Germline mosaicism of a missense variant in <scp>KCNC2</scp> in a multiplex family with autism and epilepsy characterized by longâ€read sequencing. American Journal of Medical Genetics, Part A, 2022, 188, 2071-2081.	0.7	7
3980	Ancestry adjustment improves genome-wide estimates of regional intolerance. Genetics, 2022, , .	1.2	2
3981	Exome sequencing of individuals with Huntingtonâ€™s disease implicates FAN1 nuclease activity in slowing CAG expansion and disease onset. Nature Neuroscience, 2022, 25, 446-457.	7.1	31

#	Article	IF	Citations
3982	De novo heterozygous variants in KIF5B cause kyphomelic dysplasia. Clinical Genetics, 2022, 102, 3-11.	1.0	5
3983	<scp>IQSEC2</scp>â€related encephalopathy in males due to missense variants in the pleckstrin homology domain. Clinical Genetics, 2022, 102, 72-77.	1.0	4
3984	Maternal effect genes as risk factors for congenital heart defects. Human Genetics and Genomics Advances, 2022, 3, 100098.	1.0	2
3985	A complete reference genome improves analysis of human genetic variation. Science, 2022, 376, eabl3533.	6.0	144
3986	Genome-wide analysis of somatic noncoding mutation patterns in cancer. Science, 2022, 376, eabg5601.	6.0	33
3987	AOPEP variants as a novel cause of recessive dystonia: Generalized dystonia and dystonia-parkinsonism. Parkinsonism and Related Disorders, 2022, 97, 52-56.	1.1	7
3988	Identifying Genetic Biomarkers Predicting Response to Anti-Vascular Endothelial Growth Factor Injections in Diabetic Macular Edema. International Journal of Molecular Sciences, 2022, 23, 4042.	1.8	5
3989	Bioinformatic Challenges Detecting Genetic Variation in Precision Medicine Programs. Frontiers in Medicine, 2022, 9, 806696.	1.2	3
3990	Loss-of-function variants in TIAM1 are associated with developmental delay, intellectual disability, and seizures. American Journal of Human Genetics, 2022, 109, 571-586.	2.6	19
3991	Phenotypeâ€driven approaches to enhance variant prioritization and diagnosis of rare disease. Human Mutation, 2022, 43, 1071-1081.	1.1	17
3992	Pathophysiological Heterogeneity of the BBSOA Neurodevelopmental Syndrome. Cells, 2022, 11, 1260.	1.8	10
3993	Homozygosity for a novel INHA mutation in two male siblings with hypospadias, primary hypogonadism, and high-normal testicular volume. European Journal of Endocrinology, 2022, 186, K25-K31.	1.9	2
3994	A novel nonsense variant in MED12 associated with malformations in a female fetus. Clinical Case Reports (discontinued), 2021, 9, e05124.	0.2	1
3995	Mutation screening of the DNAJC7 gene in Japanese patients with sporadic amyotrophic lateral sclerosis. Neurobiology of Aging, 2022, 113, 131-136.	1.5	6
3996	Genetic Risk in Families with Age-Related Macular Degeneration. Ophthalmology Science, 2021, 1, 100087.	1.0	5
3997	Genetic landscape of human mitochondrial genome using whole-genome sequencing. Human Molecular Genetics, 2022, 31, 1747-1761.	1.4	4
3998	Exome Sequencing Reveals Novel TTN Variants in Saudi Patients with Congenital Titinopathies. Genetic Testing and Molecular Biomarkers, 2021, 25, 757-764.	0.3	4
3999	Variant-specific effects define the phenotypic spectrum of HNRNPH2-associated neurodevelopmental disorders in males. Human Genetics, 2022, 141, 257-272.	1.8	8

#	Article	IF	Citations
4001	Precision Medicine through Next-Generation Sequencing in Inherited Eye Diseases in a Korean Cohort. Genes, 2022, 13, 27.	1.0	11
4002	Intellectual disability genomics: current state, pitfalls and future challenges. BMC Genomics, 2021, 22, 909.	1.2	31
4003	Successful outcomes of intracytoplasmic sperm injectionâ€“embryo transfer using ejaculated spermatozoa from two Chinese asthenoteratozoospermic brothers with a compound heterozygous FSIP2 mutation. Andrologia, 2022, 54, e14351.	1.0	7
4004	Can the Synergic Contribution of Multigenic Variants Explain the Clinical and Cellular Phenotypes of a Neurodevelopmental Disorder?. Genes, 2022, 13, 78.	1.0	1
4005	Screening and Functional Analysis of TEK Mutations in Chinese Children With Primary Congenital Glaucoma. Frontiers in Genetics, 2021, 12, 764509.	1.1	7
4007	Whole exome sequencing identifies deleterious rare variants in CCDC141 in familial self-limited delayed puberty. Npj Genomic Medicine, 2021, 6, 107.	1.7	4
4008	PPIL4 is essential for brain angiogenesis and implicated in intracranial aneurysms in humans. Nature Medicine, 2021, 27, 2165-2175.	15.2	23
4009	Clinical, Histological, and Genetic Features of 25 Patients with Autosomal Dominant Progressive External Ophthalmoplegia (ad-PEO)/PEO-Plus Due to TWNK Mutations. Journal of Clinical Medicine, 2022, 11, 22.	1.0	5
4010	Prioritising positively selected variants in whole-genome sequencing data using FineMAV. BMC Bioinformatics, 2021, 22, 604.	1.2	0
4011	De novo mutations in childhood cases of sudden unexplained death that disrupt intracellular Ca ²⁺ regulation. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	21
4012	A homozygous in-frame duplication within the LRRCT consensus sequence of CFAP410 causes cone-rod dystrophy, macular staphyloma and short stature. Ophthalmic Genetics, 2022, 43, 378-384.	0.5	3
4013	TM2D genes regulate Notch signaling and neuronal function in Drosophila. PLoS Genetics, 2021, 17, e1009962.	1.5	5
4016	Novel PTCH1 and concurrent TP53 mutations in four patients with numerous nonâ€syndromic basal cell carcinomas: The paradigm of oncogenic synergy. Experimental Dermatology, 2021, , .	1.4	2
4017	A full-proteome, interaction-specific characterization of mutational hotspots across human cancers. Genome Research, 2022, 32, 135-149.	2.4	2
4018	Prevalence of germline TP53 mutation among early onset middle eastern breast cancer patients. Hereditary Cancer in Clinical Practice, 2021, 19, 49.	0.6	2
4019	Prediction of disease-associated nsSNPs by integrating multi-scale ResNet models with deep feature fusion. Briefings in Bioinformatics, 2022, 23, .	3.2	13
4020	RegVar: Tissue-Specific Prioritization of Non-Coding Regulatory Variants. Genomics, Proteomics and Bioinformatics, 2023, 21, 385-395.	3.0	2
4021	Predicting deleterious missense genetic variants via integrative supervised nonnegative matrix tri-factorization. Scientific Reports, 2021, 11, 23747.	1.6	0

#	Article	IF	Citations
4022	SNPs Filtered by Allele Frequency Improve the Prediction of Hypertension Subtypes. , 2021, , .		0
4023	Embeddings from protein language models predict conservation and variant effects. Human Genetics, 2022, 141, 1629-1647.	1.8	60
4025	A rare CTSC mutation in Papillon-LefÃ¨vre Syndrome results in abolished serine protease activity and reduced NET formation but otherwise normal neutrophil function. PLoS ONE, 2021, 16, e0261724.	1.1	4
4026	The microRNA processorDROSHAis a candidate gene for a severe progressive neurological disorder. Human Molecular Genetics, 2022, 31, 2934-2950.	1.4	6
4027	The use of base editing technology to characterize single nucleotide variants. Computational and Structural Biotechnology Journal, 2022, 20, 1670-1680.	1.9	4
4029	A Severe Dementia Syndrome Caused by Intron Retention and Cryptic Splice Site Activation in STUB1 and Exacerbated by TBP Repeat Expansions. Frontiers in Molecular Neuroscience, 2022, 15, 878236.	1.4	8
4030	The Clinical Variant Analysis Tool: Analyzing the evidence supporting reported genomic variation in clinical practice. Genetics in Medicine, 2022, 24, 1512-1522.	1.1	4
4031	A Novel FGFR1 Missense Mutation in a Portuguese Family with Congenital Hypogonadotropic Hypogonadism. International Journal of Molecular Sciences, 2022, 23, 4423.	1.8	0
4032	A general framework for predicting the transcriptomic consequences of non-coding variation and small molecules. PLoS Computational Biology, 2022, 18, e1010028.	1.5	2
4033	Heterozygous <scp>PNPT1</scp> Variants Cause Spinocerebellar Ataxia Type 25. Annals of Neurology, 2022, 92, 122-137.	2.8	8
4035	Genome-Wide Association Study of Fluorescent Oxidation Products Accounting for Tobacco Smoking Status in Adults from the French EGEA Study. Antioxidants, 2022, 11, 802.	2.2	3
4037	Diagnostic yield of whole exome data in fetuses aborted for conotruncal malformations. Prenatal Diagnosis, 2022, 42, 852-861.	1.1	1
4038	Cathepsin F genetic mutation is associated with familial papillary thyroid cancer. American Journal of the Medical Sciences, 2022, 364, 414-424.	0.4	3
4039	Frequency of pathogenic germline variants in cancer susceptibility genes in 1336 renal cell carcinoma cases. Human Molecular Genetics, 2022, 31, 3001-3011.	1.4	9
4041	Whole mitochondrial genome sequencing of Malaysian patients with cardiomyopathy. PeerJ, 2022, 10, e13265.	0.9	1
4042	Contribution of Whole-Genome Sequencing and Transcript Analysis to Decipher Retinal Diseases Associated with MFSD8 Variants. International Journal of Molecular Sciences, 2022, 23, 4294.	1.8	3
4043	Phenotypic overlap between cardioacrofacial dysplasia-2 and oral-facial-digital syndrome. European Journal of Medical Genetics, 2022, , 104512.	0.7	0
4044	Severe COVID-19-associated variants linked to chemokine receptor gene control in monocytes and macrophages. Genome Biology, 2022, 23, 96.	3.8	17

#	Article	IF	Citations
4045	The Challenge of Genetic Variants of Uncertain Clinical Significance. Annals of Internal Medicine, 2022, 175, 994-1000.	2.0	29
4195	Next-generation sequencing identified novel truncating mutations in BBS9 causing Bardet Biedl syndrome in two Iranian consanguineous families.. Iranian Journal of Child Neurology, 2022, 16, 123-133.	0.2	0
4197	Pre-Implantation Genetic Testing for Monogenic Disorders (PGT-M) in A Family with A Novel Mutation in Gene. Cell Journal, 2021, 23, 593-597.	0.2	0
4198	Novel Mutation in the First Tunisian Family With Warburg Micro Syndrome.. Journal of Clinical		

#	Article	IF	Citations
4215	Genetic insight into Birtâ€“Hoggâ€“DubÃ© syndrome in Indian patients reveals novel mutations at FLCN. Orphanet Journal of Rare Diseases, 2022, 17, 176.	1.2	1
4216	OMEN: network-based driver gene identification using mutual exclusivity. Bioinformatics, 2022, 38, 3245-3251.	1.8	4
4217	Whole-exome sequencing identifies rare genetic variants associated with human plasma metabolites. American Journal of Human Genetics, 2022, 109, 1038-1054.	2.6	17
4218	Whole exome sequencing identifies novel germline variants of SLC15A4 gene as potentially cancer predisposing in familial colorectal cancer. Molecular Genetics and Genomics, 2022, , 1.	1.0	1
4219	Genome-wide association and Mendelian randomization study of blood copper levels and 213 deep phenotypes in humans. Communications Biology, 2022, 5, 405.	2.0	7
4220	Integration of Protein Structure and Population-Scale DNA Sequence Data for Disease Gene Discovery and Variant Interpretation. Annual Review of Biomedical Data Science, 2022, 5, .	2.8	0
4221	Complement Gene Variant Effect on Relapse of Complement-Mediated Thrombotic Microangiopathy after Eculizumab Cessation. Blood Advances, 2022, , .	2.5	2
4222	De novo variants in the PABP domain of PABPC1 lead to developmental delay. Genetics in Medicine, 2022, , .	1.1	4
4223	RNA-Based Targeted Gene Sequencing Improves the Diagnostic Yield of Mutant Detection in Chronic Myeloid Leukemia. Journal of Molecular Diagnostics, 2022, 24, 803-822.	1.2	2
4224	VWFâ€Gly2752Ser, a novel nonâ€cysteine substitution variant in the CK domain, exhibits severe secretory impairment by hampering Câ€terminal dimer formation. Journal of Thrombosis and Haemostasis, 2022, , .	1.9	1
4225	Rare variants implicate NMDA receptor signaling and cerebellar gene networks in risk for bipolar disorder. Molecular Psychiatry, 2022, 27, 3842-3856.	4.1	5
4226	Shared genetic loci between depression and cardiometabolic traits. PLoS Genetics, 2022, 18, e1010161.	1.5	18
4227	Analysis of MRI-derived spleen iron in the UK Biobank identifies genetic variation linked to iron homeostasis and hemolysis. American Journal of Human Genetics, 2022, 109, 1092-1104.	2.6	7
4228	Massively parallel identification of functionally consequential noncoding genetic variants in undiagnosed rare disease patients. Scientific Reports, 2022, 12, 7576.	1.6	1
4229	Recombination affects allele-specific expression of deleterious variants in human populations. Science Advances, 2022, 8, eabl3819.	4.7	3
4230	Contribution of rare whole-genome sequencing variants to plasma protein levels and the missing heritability. Nature Communications, 2022, 13, 2532.	5.8	9
4231	Further description of two patients with biallelic variants in <scp>NADSYN1</scp> in association with cardiac and vertebral anomalies. American Journal of Medical Genetics, Part A, 2022, 188, 2479-2484.	0.7	8
4232	Rare Variant Analysis and Molecular Dynamics Simulation in Alzheimerâ€™s Disease Identifies Exonic Variants in FLG. Genes, 2022, 13, 838.	1.0	6

#	Article	IF	Citations
4233	Protposer: The web server that readily proposes protein stabilizing mutations with high PPV. Computational and Structural Biotechnology Journal, 2022, 20, 2415-2433.	1.9	3
4234	A De Novo Missense <scp>NPTX1</scp> Variant in an Individual with Infantileâ€Onset Cerebellar Ataxia. Movement Disorders, 2022, 37, 1774-1776.	2.2	1
4235	Gene-based whole genome sequencing meta-analysis of 250 circulating proteins in three isolated European populations. Molecular Metabolism, 2022, 61, 101509.	3.0	3
4236	Characterizing the polygenic overlaps of bipolar disorder subtypes with schizophrenia and major depressive disorder. Journal of Affective Disorders, 2022, 309, 242-251.	2.0	3
4237	GIMAP6 regulates autophagy, immune competence, and inflammation in mice and humans. Journal of Experimental Medicine, 2022, 219, .	4.2	4
4238	Whole exome sequencing of known eye genes reveals genetic causes for high myopia. Human Molecular Genetics, 2022, 31, 3290-3298.	1.4	16
4239	Opportunities and challenges for the use of common controls in sequencing studies. Nature Reviews Genetics, 2022, 23, 665-679.	7.7	13
4240	Complex Presentation of Hao-Fountain Syndrome Solved by Exome Sequencing Highlighting Co-Occurring Genomic Variants. Genes, 2022, 13, 889.	1.0	7
4241	Genome-wide association meta-analysis identifies 48 risk variants and highlights the role of the stria vascularis in hearing loss. American Journal of Human Genetics, 2022, 109, 1077-1091.	2.6	27
4242	Evaluation of cfDNA as an early detection assay for dense tissue breast cancer. Scientific Reports, 2022, 12, 8458.	1.6	3
4243	Computational and experimental methods for classifying variants of unknown clinical significance.. Cold Spring Harbor Molecular Case Studies, 2022, 8, .	0.7	7
4244	Human OTULIN haploinsufficiency impairs cell-intrinsic immunity to staphylococcal Î±-toxin. Science, 2022, 376, eabm6380.	6.0	25
4245	Breast cancer risks associated with missense variants in breast cancer susceptibility genes. Genome Medicine, 2022, 14, 51.	3.6	19
4247	Innovative in Silico Approaches for Characterization of Genes and Proteins. Frontiers in Genetics, 2022, 13, .	1.1	6
4249	A Brief Review on Deep Learning Applications in Genomic Studies. Frontiers in Systems Biology, 2022, 2, .	0.5	7
4251	Identification of a Complex Allele in IMPG2 as a Cause of Adult-Onset Vitelliform Macular Dystrophy. , 2022, 63, 27.		13
4253	Integrative Approach to Predict Severity in Nonketotic Hyperglycinemia. Annals of Neurology, 2022, 92, 292-303.	2.8	3
4254	Benchmarking of univariate pleiotropy detection methods applied to epilepsy. Human Mutation, 2022, 43, 1314-1332.	1.1	0

#	Article	IF	Citations
4257	A Study of the Genomic Variations Associated with Autistic Spectrum Disorders in a Russian Cohort of Patients Using Whole-Exome Sequencing. Genes, 2022, 13, 920.	1.0	3
4258	Rare Germline Variants Are Associated with Rapid Biochemical Recurrence After Radical Prostate Cancer Treatment: A Pan Prostate Cancer Group Study. European Urology, 2022, 82, 201-211.	0.9	2
4259	Defining mitochondrial protein functions through deep multiomic profiling. Nature, 2022, 606, 382-388.	13.7	49
4260	Analysis of Mutational Profile of Hypopharyngeal and Laryngeal Head and Neck Squamous Cell Carcinomas Identifies KMT2C as a Potential Tumor Suppressor. Frontiers in Oncology, 0, 12, .	1.3	3
4261	Cognitive Impairment in a Complex Family With AAGGG and ACAGG Repeat Expansions in RFC1 Detected by ExpansionHunter Denovo. Neurology: Genetics, 2022, 8, .	0.9	4
4262	Description of Two New Cases of AQP1 Related Pulmonary Arterial Hypertension and Review of the Literature. Genes, 2022, 13, 927.	1.0	5
4263	Diverse molecular causes of unsolved autosomal dominant tubulointerstitial kidney diseases. Kidney International, 2022, 102, 405-420.	2.6	10
4264	Rare Variants in Inborn Errors of Immunity Genes Associated With Covid-19 Severity. Frontiers in Cellular and Infection Microbiology, 2022, 12, .	1.8	5
4268	Identification of PSMB5 as a genetic modifier of fragile Xâ€“associated tremor/ataxia syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	3.3	7
4269	Omega-3 fatty acid epoxides produced by PAF-AH2 in mast cells regulate pulmonary vascular remodeling. Nature Communications, 2022, 13, .	5.8	13
4271	Protein Network Analysis of Whole Exome Sequencing of Severe Preeclampsia. Frontiers in Genetics, 2022, 12, .	1.1	2
4272	New Genetic Variants in CYP2B6 and SLC6A Support the Role of Oxidative Stress in Familial MÃ©niÃ¨reâ€™s Disease. Genes, 2022, 13, 998.	1.0	5
4273	Quantitative assessment of copy number alterations by liquid biopsy for neuroblastoma. Genes Chromosomes and Cancer, 2022, 61, 662-669.	1.5	6
4274	Additive genetic effect of GCKR, G6PC2, and SLC30A8 variants on fasting glucose levels and risk of type 2 diabetes. PLoS ONE, 2022, 17, e0269378.	1.1	4
4277	PON-All: Amino Acid Substitution Tolerance Predictor for All Organisms. Frontiers in Molecular Biosciences, 0, 9, .	1.6	7
4279	A diseaseâ€associated missense mutation in CYP4F3 affects the metabolism of leukotriene B4 via disruption of electron transfer. Journal of Cachexia, Sarcopenia and Muscle, 2022, 13, 2242-2253.	2.9	5
4281	Exome sequencing identifies genetic variants in anophthalmia and microphthalmia. American Journal of Medical Genetics, Part A, 2022, 188, 2376-2388.	0.7	2
4282	Interpreting protein variant effects with computational predictors and deep mutational scanning. DMM Disease Models and Mechanisms, 2022, 15, .	1.2	25

#	Article	IF	Citations
4283	TTC5 syndrome: Clinical and molecular spectrum of a severe and recognizable condition. American Journal of Medical Genetics, Part A, 2022, 188, 2652-2665.	0.7	4
4284	A Novel Somatic Mutation of CACNA1H p.V1937M in Unilateral Primary Hyperaldosteronism. Frontiers in Endocrinology, 0, 13, .	1.5	2
4285	Distributed genetic architecture across the hippocampal formation implies common neuropathology across brain disorders. Nature Communications, 2022, 13, .	5.8	12
4287	Artificial intelligence and machine learning in precision and genomic medicine. , 2022, 39, .		53
4288	New Developments and Possibilities in Reanalysis and Reinterpretation of Whole Exome Sequencing Datasets for Unsolved Rare Diseases Using Machine Learning Approaches. International Journal of Molecular Sciences, 2022, 23, 6792.	1.8	9
4289	Molecular deciphering of primary liver neuroendocrine neoplasms confirms their distinct existence with foregutâ€like profile. Journal of Pathology, 2022, 258, 58-68.	2.1	3
4290	Functional characterization of a novel p.Ser76Thr variant in IGFBP4 that associates with body mass index in American Indians. European Journal of Human Genetics, 0, , .	1.4	1
4291	Exome sequencing in multiplex families with left-sided cardiac defects has high yield for disease gene discovery. PLoS Genetics, 2022, 18, e1010236.	1.5	8
4292	PTBP2 â€“ a gene with relevance for both Anorexia nervosa and body weight regulation. Translational Psychiatry, 2022, 12, .	2.4	4
4293	TSABL: Trait Specific Annotation Based Locus predictor. BMC Genomics, 2022, 23, .	1.2	0
4295	Single-cell characterization of step-wise acquisition of carboplatin resistance in ovarian cancer. Npj Systems Biology and Applications, 2022, 8, .	1.4	2
4296	Loss-of-function variant in chymotrypsin like elastase 3B (CELA3B) is associated with non-alcoholic chronic pancreatitis. Pancreatology, 2022, 22, 713-718.	0.5	8
4297	Human rs75776403 polymorphism links differential phenotypic and clinical outcomes to a CLEC18A p.T151M-driven multiomics. Journal of Biomedical Science, 2022, 29, .	2.6	0
4298	Impact of Genetic Testing on Therapeutic Decision-Making in Childhood-Onset Epilepsiesâ€”a Study in a Tertiary Epilepsy Center. Neurotherapeutics, 2022, 19, 1353-1367.	2.1	14
4299	Respiratory viral infections in otherwise healthy humans with inherited IRF7 deficiency. Journal of Experimental Medicine, 2022, 219, .	4.2	21
4301	scEpiLock: A Weakly Supervised Learning Framework for cis-Regulatory Element Localization and Variant Impact Quantification for Single-Cell Epigenetic Data. Biomolecules, 2022, 12, 874.	1.8	0
4302	Causal Inference of Genetic Variants and Genes in Amyotrophic Lateral Sclerosis. Frontiers in Genetics, 0, 13, .	1.1	4
4303	Integrating variant functional annotation scores have varied abilities to improve power of genome-wide association studies. Scientific Reports, 2022, 12, .	1.6	1

#	Article	IF	Citations
4304	Immunophenotypic and molecular characterization of pancreatic neuroendocrine tumors producing serotonin. Modern Pathology, 2022, 35, 1713-1722.	2.9	4
4305	Possible association of 16p11.2 copy number variation with altered lymphocyte and neutrophil counts. Npj Genomic Medicine, 2022, 7, .	1.7	3
4306	Missense variants causing Wiedemann-Steiner syndrome preferentially occur in the KMT2A-CXXC domain and are accurately classified using AlphaFold2. PLoS Genetics, 2022, 18, e1010278.	1.5	3
4307	Pinpointing novel risk loci for Lewy body dementia and the shared genetic etiology with Alzheimerâ€™s disease and Parkinsonâ€™s disease: a large-scale multi-trait association analysis. BMC Medicine, 2022, 20, .	2.3	10
4308	Drug screen in iPSC-Neurons identifies nucleoside analogs as inhibitors of (G4C2)n expression in C9orf72 ALS/FTD. Cell Reports, 2022, 39, 110913.	2.9	7
4311	An accurate prediction model of digenic interaction for estimating pathogenic gene pairs of human diseases. Computational and Structural Biotechnology Journal, 2022, 20, 3639-3652.	1.9	4
4312	dbBIP: a comprehensive bipolar disorder database for genetic research. Database: the Journal of Biological Databases and Curation, 2022, 2022, .	1.4	3
4313	Whole-exome sequencing deciphers the genetic profile of visual impairments in patients from Southwest Iran. Molecular Genetics and Genomics, 0, , .	1.0	1
4314	The Location of Missense Variants in the Human GIP Gene Is Indicative for Natural Selection. Frontiers in Endocrinology, 0, 13, .	1.5	1
4315	Proactive functional classification of all possible missense single-nucleotide variants in KCNQ4. Genome Research, 2022, 32, 1573-1584.	2.4	8
4316	Polygenic risk score for ACE-inhibitor-associated cough based on the discovery of new genetic loci. European Heart Journal, 2022, 43, 4707-4718.	1.0	5
4317	Networkâ€based metaâ€analysis and the candidate gene association studies reveal novel ethnicityâ€specific variants in <scp> MFSD3 </scp> and <scp> MRPL43 </scp> associated with dementia with Lewy bodies. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 0, , .	1.1	1
4318	De novo heterozygous variants in <scp>SLC30A7</scp> are a candidate cause for Joubert syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 2360-2366.	0.7	3
4320	Genetic Relationships between Attention-Deficit/Hyperactivity Disorder, Autism Spectrum Disorder, and Intelligence. Neuropsychobiology, 2022, 81, 484-496.	0.9	26
4321	A novel mutation in GJC2 associated with hypomyelinating leukodystrophy type 2 disorder. Genomics and Informatics, 2022, 20, e24.	0.4	1
4323	Case Report: A Novel Homozygous Missense Variant of FBN3 Supporting It Is a New Candidate Gene Causative of a Bardetâ€“Biedl Syndromeâ€“Like Phenotype. Frontiers in Genetics, 0, 13, .	1.1	1
4325	Differences in actionable genomic alterations between brain metastases and nonâ€‘brain metastases in patients with nonâ€‘small cell lung cancer. International Journal of Oncology, 2022, 61, .	1.4	1
4326	PRENATAL ULTRASOUND FINDINGS ASSOCIATED WITH PIGW VARIANTS:: ONE MORE PIECE IN THE FRYNS SYNDROME PUZZLE? PIGW â€related prenatal findings. Prenatal Diagnosis, 0, , .	1.1	1

#	Article	IF	Citations
4327	Case Report: Primary Leptomeningeal Medulloblastoma in a Child: Clinical Case Report and Literature Review. Frontiers in Pediatrics, 0, 10, .	0.9	1
4329	No evidence of increased mutations in the germline of a group of British nuclear test veterans. Scientific Reports, 2022, 12, .	1.6	6
4330	Large scale population screening for Duchenne muscular dystrophyâ€"Predictable and unpredictable challenges. Prenatal Diagnosis, 2022, 42, 1162-1172.	1.1	2
4331	The Prevalence and Genetic Spectrum of Familial Hypercholesterolemia in Qatar Based on Whole Genome Sequencing of 14,000 Subjects. Frontiers in Genetics, 0, 13, .	1.1	1
4333	Large-scale analyses of the X chromosome in 2,354 infertile men discover recurrently affected genes associated with spermatogenic failure. American Journal of Human Genetics, 2022, 109, 1458-1471.	2.6	10
4334	Uncovering the Contribution of Moderate-Penetrance Susceptibility Genes to Breast Cancer by Whole-Exome Sequencing and Targeted Enrichment Sequencing of Candidate Genes in Women of European Ancestry. Cancers, 2022, 14, 3363.	1.7	2
4335	CaMKII Inhibition Attenuates Distinct Gain-of-Function Effects Produced by Mutant Nav1.6 Channels and Reduces Neuronal Excitability. Cells, 2022, 11, 2108.	1.8	2
4336	Genome-wide meta-analysis of insomnia prioritizes genes associated with metabolic and psychiatric pathways. Nature Genetics, 2022, 54, 1125-1132.	9.4	61
4338	LmTag: functional-enrichment and imputation-aware tag SNP selection for population-specific genotyping arrays. Briefings in Bioinformatics, 2022, 23, .	3.2	1
4339	The evolutionary history of human spindle genes includes back-and-forth gene flow with Neandertals. ELife, 0, 11, .	2.8	12
4340	SLITRK2 variants associated with neurodevelopmental disorders impair excitatory synaptic function and cognition in mice. Nature Communications, 2022, 13, .	5.8	6
4341	Genome-wide association study of cerebellar volume provides insights into heritable mechanisms underlying brain development and mental health. Communications Biology, 2022, 5, .	2.0	3
4342	Association of mitochondrial DNA content, heteroplasmies and inter-generational transmission with autism. Nature Communications, 2022, 13, .	5.8	14
4343	The Phenotypic Continuum of ATP1A3 -Related Disorders. Neurology, 2022, 99, .	1.5	16
4344	The role of single-cell genomics in human genetics. Journal of Medical Genetics, 2022, 59, 827-839.	1.5	11
4345	Non-coding de novo mutations in chromatin interactions are implicated in autism spectrum disorder. Molecular Psychiatry, 2022, 27, 4680-4694.	4.1	9
4346	Genome-wide association meta-analysis of 88,250 individuals highlights pleiotropic mechanisms of five ocular diseases in UK Biobank. EBioMedicine, 2022, 82, 104161.	2.7	21
4347	Variant Annotation and Functional Prediction: SnpEff. Methods in Molecular Biology, 2022, , 289-314.	0.4	47

#	Article	IF	Citations
4348	Whole genome sequencing and inheritance-based variant filtering as a tool for unraveling missing heritability in pediatric cancer. Pediatric Hematology and Oncology, 2023, 40, 326-340.	0.3	0
4349	Ras-mutant cancers are sensitive to small molecule inhibition of V-type ATPases in mice. Nature Biotechnology, 2022, 40, 1834-1844.	9.4	13
4350	Investigation of CACNA1I Cav3.3 Dysfunction in Hemiplegic Migraine. Frontiers in Molecular Neuroscience, 0, 15, .	1.4	7
4351	Autosomal dominant optic atrophy caused by six novel pathogenic OPA1 variants and genotypeâ€“phenotype correlation analysis. BMC Ophthalmology, 2022, 22, .	0.6	1
4352	Unfolding the genotype-to-phenotype black box of cardiovascular diseases through cross-scale modeling. IScience, 2022, 25, 104790.	1.9	1
4353	Novel RAB27A Variant Associated with Late-Onset Hemophagocytic Lymphohistiocytosis Alters Effector Protein Binding. Journal of Clinical Immunology, 2022, 42, 1685-1695.	2.0	1
4354	FUS-P525L Juvenile Amyotrophic Lateral Sclerosis and Intellectual Disability. Neurology: Genetics, 2022, 8, .	0.9	2
4355	Integrated Quantile RAnk Test (iQRAT) for gene-level associations. Annals of Applied Statistics, 2022, 16, .	0.5	3
4356	Using Machine Learning for Predicting the Effect of Mutations in the Initiation Codon. IEEE Journal of Biomedical and Health Informatics, 2022, 26, 5750-5756.	3.9	1
4358	Identification of RNF13 as cause of recessively inherited ALS in a multi-case pedigree. Journal of Medical Genetics, 0, , jmedgenet-2022-108645.	1.5	1
4359	Mutational Profile and Potential Molecular Therapeutic Targets of Pheochromocytoma. Frontiers in Endocrinology, 0, 13, .	1.5	1
4360	Targeted re-sequencing on 1p22 among non-syndromic orofacial clefts from Han Chinese population. Frontiers in Genetics, 0, 13, .	1.1	0
4361	Disease Progression of WHIM Syndrome in an International Cohort of 66 Pediatric and Adult Patients. Journal of Clinical Immunology, 2022, 42, 1748-1765.	2.0	9
4363	Prediction of post-translational modification cross-talk and mutation within proteins via imbalanced learning. Expert Systems With Applications, 2023, 211, 118593.	4.4	0
4365	Demystifying non-coding GWAS variants: an overview of computational tools and methods. Human Molecular Genetics, 2022, 31, R73-R83.	1.4	9
4367	Dominantly acting KIF5B variants with pleiotropic cellular consequences cause variable clinical phenotypes. Human Molecular Genetics, 2023, 32, 473-488.	1.4	4
4369	High-throughput sequencing analysis of nuclear-encoded mitochondrial genes reveals a genetic signature of human longevity. GeroScience, 2023, 45, 311-330.	2.1	5
4370	Genome-wide meta-analyses reveal novel loci for verbal short-term memory and learning. Molecular Psychiatry, 2022, 27, 4419-4431.	4.1	6

#	Article	IF	Citations
4371	Evidence of a genetically driven metabolomic signature in actively inflamed Crohnâ€™s disease. Scientific Reports, 2022, 12, .	1.6	3
4372	GWAS meta-analysis of intrahepatic cholestasis of pregnancy implicates multiple hepatic genes and regulatory elements. Nature Communications, 2022, 13, .	5.8	11
4373	A paradigm shift in pharmacogenomics: From candidate polymorphisms to comprehensive sequencing. Basic and Clinical Pharmacology and Toxicology, 0, , .	1.2	6
4374	Variant rs4149584 (R92Q) of the TNFRSF1A gene in patients with familial multiple sclerosis. NeurologÃa (English Edition), 2022, , .	0.2	1
4375	Genetic predisposition to portoâ€sinusoidal vascular disorder: A functional genomicâ€based, multigenerational family study. Hepatology, 2023, 77, 501-511.	3.6	7
4376	Whole Exome Sequencing Revealed Variants That Predict Pulmonary Artery Involvement in Patients with Takayasu Arteritis. Journal of Inflammation Research, 0, Volume 15, 4817-4831.	1.6	3
4377	Clinical and genetic characterization of NEFL-related neuropathy in Taiwan. Journal of the Formosan Medical Association, 2022, , .	0.8	2
4379	KBG syndrome: videoconferencing and use of artificial intelligence driven facial phenotyping in 25 new patients. European Journal of Human Genetics, 2022, 30, 1244-1254.	1.4	13
4381	Functional regulatory variants implicate distinct transcriptional networks in dementia. Science, 2022, 377, .	6.0	49
4382	AutoCaSc: Prioritizing candidate genes for neurodevelopmental disorders. Human Mutation, 2022, 43, 1795-1807.	1.1	5
4383	Targeted Sequencing of Plasma-Derived vs. Urinary cfDNA from Patients with Triple-Negative Breast Cancer. Cancers, 2022, 14, 4101.	1.7	5
4384	Exploring Lead loci shared between schizophrenia and Cardiometabolic traits. BMC Genomics, 2022, 23, .	1.2	1
4385	Pathogenic genetic variants identified in Australian families with paediatric cataract. BMJ Open Ophthalmology, 2022, 7, e001064.	0.8	7
4386	Genetic variants with discordant classifications: An assessment of genetic counselor attitudes and practices. Journal of Genetic Counseling, 0, , .	0.9	1
4387	Cancer-driving mutations are enriched in genic regions intolerant to germline variation. Science Advances, 2022, 8, .	4.7	2
4389	Whole exome and genome sequencing in mendelian disorders: a diagnostic and health economic analysis. European Journal of Human Genetics, 2022, 30, 1121-1131.	1.4	34
4390	Germline Missense Variants in CDC20 Result in Aberrant Mitotic Progression and Familial Cancer. Cancer Research, 2022, 82, 3499-3515.	0.4	4
4391	The secondary pocket of cryptochrome 2 is important for the regulation of its stability and localization. Journal of Biological Chemistry, 2022, 298, 102334.	1.6	5

#	Article	IF	Citations
4392	Clinical presentation and molecular genetic analysis of a Sudanese family with a novel mutation in the CYP2R1 gene. Gene, 2022, 844, 146809.	1.0	1
4394	The recurrent de novo c.2011C>T missense variant in MTSS2 causes syndromic intellectual disability. American Journal of Human Genetics, 2022, 109, 1923-1931.	2.6	7
4395	A Novel Nonsense Variant in GRM1 Causes Autosomal Recessive Spinocerebellar Ataxia 13 in a Consanguineous Pakistani Family. Genes, 2022, 13, 1667.	1.0	7
4396	Challenges in quantifying genome erosion for conservation. Frontiers in Genetics, 0, 13, .	1.1	6
4397	Systems analysis of de novo mutations in congenital heart diseases identified a protein network in the hypoplastic left heart syndrome. Cell Systems, 2022, 13, 895-910.e4.	2.9	3
4398	Genetic variability shapes the alternative pathway complement activity and predisposition to complementâ€related diseases. Immunological Reviews, 2023, 313, 71-90.	2.8	15
4399	TRIT1 deficiency: Two novel patients with four novel variants. European Journal of Medical Genetics, 2022, 65, 104603.	0.7	1
4400	Challenges Related to the Use of Next-Generation Sequencing for the Optimization of Drug Therapy. Handbook of Experimental Pharmacology, 2022, , .	0.9	0
4401	An Examination of Mechanisms by which Synonymous Mutations may Alter Protein Levels, Structure and Functions. , 2022, , 99-132.		1
4402	Using Genome Wide Studies to Generate and Test Hypotheses that Provide Mechanistic Details of How Synonymous Codons Affect Protein Structure and Function: Functional SNPs in the Age of Precision Medicine. , 2022, , 171-183.		0
4403	SNPs and Personalized Medicine: Scrutinizing Pathogenic Synonymous Mutations for Precision Oncology. , 2022, , 185-195.		0
4404	MAB21L4 Deficiency Drives Squamous Cell Carcinoma via Activation of RET. Cancer Research, 2022, 82, 3143-3157.	0.4	3
4405	In silico analysis of upstream variants in Brazilian patients with Familial hypercholesterolemia. Gene, 2023, 849, 146908.	1.0	2
4407	Identifying novel genetic loci associated with polycystic ovary syndrome based on its shared genetic architecture with type 2 diabetes. Frontiers in Genetics, 0, 13, .	1.1	1
4408	Mutations in BCOR , a co-repressor of CRX/OTX2 , are associated with early-onset retinal degeneration. Science Advances, 2022, 8, .	4.7	6
4410	A Gly684Ala substitution in the androgen receptor is the cause for azoospermia in a Chinese family with mild androgen insensitivity syndrome and normal hormone levels. Frontiers in Genetics, 0, 13, .	1.1	1
4411	Identification and in-silico characterization of splice-site variants from a large cardiogenetic national registry. European Journal of Human Genetics, 2023, 31, 512-520.	1.4	3
4412	TVAR: assessing tissue-specific functional effects of non-coding variants with deep learning. Bioinformatics, 2022, 38, 4697-4704.	1.8	4

#	Article	IF	Citations
4413	A heterozygous LAMA5 variant may contribute to slowly progressive, vinculin-enhanced familial FSGS and pulmonary defects. JCI Insight, 2022, 7, .	2.3	1
4414	ATP6V0C variants impair V-ATPase function causing a neurodevelopmental disorder often associated with epilepsy. Brain, 2023, 146, 1357-1372.	3.7	6
4415	A catalog of the genetic causes of hereditary angioedema in the Canary Islands (Spain). Frontiers in Immunology, 0, 13, .	2.2	2
4416	Radiogenomic markers enable risk stratification and inference of mutational pathway states in head and neck cancer. European Journal of Nuclear Medicine and Molecular Imaging, 2023, 50, 546-558.	3.3	3
4417	Interpreting the spectrum of gamma-secretase complex missense variation in the context of hidradenitis suppurativaâ€"An in-silico study. Frontiers in Genetics, 0, 13, .	1.1	9
4418	Scalable Functional Assays for the Interpretation of Human Genetic Variation. Annual Review of Genetics, 2022, 56, 441-465.	3.2	18
4420	Dementia-related genetic variants in an Italian population of early-onset Alzheimerâ€™s disease. Frontiers in Aging Neuroscience, 0, 14, .	1.7	5
4421	A novel loss-of-function mutation of the voltage-gated potassium channel Kv10.2 involved in epilepsy and autism. Orphanet Journal of Rare Diseases, 2022, 17, .	1.2	1
4423	Genome-wide association analyses of physical activity and sedentary behavior provide insights into underlying mechanisms and roles in disease prevention. Nature Genetics, 2022, 54, 1332-1344.	9.4	64
4424	Recurrent 17q12 microduplications contribute to renal disease but not diabetes. Journal of Medical Genetics, 2023, 60, 491-497.	1.5	1
4425	Shared genetic architecture between schizophrenia and subcortical brain volumes implicates early neurodevelopmental processes and brain development in childhood. Molecular Psychiatry, 2022, 27, 5167-5176.	4.1	13
4426	The schizophrenia-associated missense variant rs13107325 regulates dendritic spine density. Translational Psychiatry, 2022, 12, .	2.4	4
4427	Genetic analyses of the electrocardiographic QT interval and its components identify additional loci and pathways. Nature Communications, 2022, 13, .	5.8	15
4428	De Novo Missense Variants in <scp>SLC32A1</scp> Cause a Developmental and Epileptic Encephalopathy Due to Impaired <scp>GABAergic</scp> Neurotransmission. Annals of Neurology, 2022, 92, 958-973.	2.8	6
4429	Ancestry-related distribution of Runs of homozygosity and functional variants in Qatari population. BMC Genomic Data, 2022, 23, .	0.7	0
4430	Novel clinical, molecular and bioinformatics insights into the genetic background of autism. Human Genomics, 2022, 16, .	1.4	2
4431	Prediction of evolutionary constraint by genomic annotations improves functional prioritization of genomic variants in maize. Genome Biology, 2022, 23, .	3.8	7
4432	Functional characteristics of a broad spectrum of TBX6Âvariants in Mayer-Rokitansky-KÃ¼ster-Hauser syndrome. Genetics in Medicine, 2022, 24, 2262-2273.	1.1	1

#	Article	IF	Citations
4433	Whole Exome Sequencing in Multi-Incident Families Identifies Novel Candidate Genes for Multiple Sclerosis. International Journal of Molecular Sciences, 2022, 23, 11461.	1.8	3
4434	Interpretation of the role of germline and somatic non-coding mutations in cancer: expression and chromatin conformation informed analysis. Clinical Epigenetics, 2022, 14, .	1.8	3
4435	Computational approaches for predicting variant impact: An overview from resources, principles to applications. Frontiers in Genetics, 0, 13, .	1.1	11
4436	Heterozygous variants in the DVL2 interaction region of DACT1 cause CAKUT and features of Townesâ€“Brocks syndrome 2. Human Genetics, 2023, 142, 73-88.	1.8	2
4437	Roles of neuroligins in central nervous system development: focus on glial neuroligins and neuron neuroligins. Journal of Translational Medicine, 2022, 20, .	1.8	9
4438	Pedigree-based study to identify GOLGB1 as a risk gene for bipolar disorder. Translational Psychiatry, 2022, 12, .	2.4	0
4440	Long-Surviving Adult Siblings With Joubert Syndrome Harboring a Novel Compound Heterozygous CPLANE1 Variant. Neurology: Genetics, 2022, 8, .	0.9	1
4443	Identification of a novel de novo pathogenic variant in GFAP in an Iranian family with Alexander disease by whole-exome sequencing. European Journal of Medical Research, 2022, 27, .	0.9	0
4444	Identification of variants in genes associated with autoinflammatory disorders in a cohort of patients with psoriatic arthritis. RMD Open, 2022, 8, e002561.	1.8	7
4445	High-coverage whole-genome sequencing of the expanded 1000 Genomes Project cohort including 602 trios. Cell, 2022, 185, 3426-3440.e19.	13.5	285
4447	DPP9 deficiency: An inflammasomopathy that can be rescued by lowering NLRP1/IL-1 signaling. Science Immunology, 2022, 7, .	5.6	13
4448	Assessing the Pathogenicity of In-Frame CACNA1F Indel Variants Using Structural Modeling. Journal of Molecular Diagnostics, 2022, 24, 1232-1239.	1.2	2
4450	A novel homozygous variant in homologous recombination repair gene ZSWIM7 causes azoospermia in males and primary ovarian insufficiency in females. European Journal of Medical Genetics, 2022, 65, 104629.	0.7	3
4451	Exome sequencing in a Swedish family with PMS2 mutation with varying penetrance of colorectal cancer: investigating the presence of genetic risk modifiers in colorectal cancer risk. European Journal of Cancer Prevention, 2023, 32, 113-118.	0.6	2
4452	Broad-scale variation in human genetic diversity levels is predicted by purifying selection on coding and non-coding elements. ELife, 0, 12, .	2.8	12
4453	Inherited IRAK-4 Deficiency in Acute Human Herpesvirus-6 Encephalitis. Journal of Clinical Immunology, 2023, 43, 192-205.	2.0	5
4454	Genome-wide study on 72,298 individuals in Korean biobank data for 76 traits. Cell Genomics, 2022, 2, 100189.	3.0	8
4455	Screening of candidate genes at GLC3B and GLC3C loci in Chinese primary congenital glaucoma patients with targeted next generation sequencing. Ophthalmic Genetics, 2023, 44, 133-138.	0.5	1

#	Article	IF	Citations
4456	A whole genome sequencing approach to anterior cruciate ligament ruptureâ€“a twin study in two unrelated families. PLoS ONE, 2022, 17, e0274354.	1.1	1
4457	NOS1 mutations cause hypogonadotropic hypogonadism with sensory and cognitive deficits that can be reversed in infantile mice. Science Translational Medicine, 2022, 14, .	5.8	15
4458	Clinical features and pathogenicity assessment in patients with HTRA1-autosomal dominant disease. Neurological Sciences, 2023, 44, 639-647.	0.9	1
4459	DeepPerVar: a multi-modal deep learning framework for functional interpretation of genetic variants in personal genome. Bioinformatics, 2022, 38, 5340-5351.	1.8	4
4461	Heterozygous pathogenic variants involvingCBFBcause a new skeletal disorder resembling cleidocranial dysplasia. Journal of Medical Genetics, 2023, 60, 498-504.	1.5	0
4463	The Notch1/CD22 signaling axis disrupts Treg function in SARS-CoV-2â€“associated multisystem inflammatory syndrome in children. Journal of Clinical Investigation, 2023, 133, .	3.9	9
4464	Cluster Analysis of Short Sensory Profile Data Reveals Sensory-Based Subgroups in Autism Spectrum Disorder. International Journal of Molecular Sciences, 2022, 23, 13030.	1.8	3
4465	The PINK1 p.Asn521Thr Variant Is Associated with Earlier Disease Onset in GRN/C9orf72 Frontotemporal Lobar Degeneration. International Journal of Molecular Sciences, 2022, 23, 12847.	1.8	1
4466	Exploration of Tools for the Interpretation of Human Non-Coding Variants. International Journal of Molecular Sciences, 2022, 23, 12977.	1.8	5
4467	Channel HCN4 mutation R666Q associated with sporadic arrhythmia decreases channel electrophysiological function and increases protein degradation. Journal of Biological Chemistry, 2022, 298, 102599.	1.6	1
4468	Identity-by-descent analysis of a large Touretteâ€™s syndrome pedigree from Costa Rica implicates genes involved in neuronal development and signal transduction. Molecular Psychiatry, 2022, 27, 5020-5027.	4.1	0
4469	The genomic landscape across 474 surgically accessible epileptogenic human brain lesions. Brain, 2023, 146, 1342-1356.	3.7	21
4472	Identification of potential driver mutations in glioblastoma using machine learning. Briefings in Bioinformatics, 2022, 23, .	3.2	9
4473	Autosomal recessive progeroid syndrome due to homozygosity for a TOMM7 variant. Journal of Clinical Investigation, 2022, 132, .	3.9	7
4474	Genomic characteristics of two breast malignant phyllodes tumors during pregnancy and lactation identified through whole-exome sequencing. Orphanet Journal of Rare Diseases, 2022, 17, .	1.2	1
4475	Association of COL12A1 rs970547 Polymorphism with Elite Athlete Status. Biomedicines, 2022, 10, 2495.	1.4	3
4476	Discovery of 42 genome-wide significant loci associated with dyslexia. Nature Genetics, 2022, 54, 1621-1629.	9.4	40
4477	Using single molecule Molecular Inversion Probes as a costâ€effective, highâ€throughput sequencing approach to target all genes and loci associated with macular diseases. Human Mutation, 2022, 43, 2234-2250.	1.1	9

#	Article	IF	Citations
4479	Specifications of the ACMG/AMP variant curation guidelines for myocilin: Recommendations from the clingen glaucoma expert panel. Human Mutation, 2022, 43, 2170-2186.	1.1	11
4482	Genetic Analysis of RASD1 as a Candidate Gene for Schizophrenia. Balkan Medical Journal, 2022, 39, 422-428.	0.3	1
4483	MicroRNA binding site variation is enriched in psychiatric disorders. Human Mutation, 2022, 43, 2153-2169.	1.1	4
4484	Identification of Genomic Variants Causing Variation in Quantitative Traits: A Review. Agriculture (Switzerland), 2022, 12, 1713.	1.4	8
4485	A framework for detecting noncoding rare-variant associations of large-scale whole-genome sequencing studies. Nature Methods, 2022, 19, 1599-1611.	9.0	36
4486	Delineation of the clinical profile of CNOT2 haploinsufficiency and overview of the <scp>IDNADFS</scp> phenotype. Clinical Genetics, 0, , .	1.0	2
4488	FGF20 and PGM2 variants are associated with childhood asthma in family-based whole-genome sequencing studies. Human Molecular Genetics, 0, , .	1.4	1
4489	Landscape of MicroRNA Regulatory Network Architecture and Functional Rerouting in Cancer. Cancer Research, 2023, 83, 59-73.	0.4	2
4490	Biallelic variants in PIGN cause Fryns syndrome, multiple congenital anomalies-hypotonia-seizures syndrome, and neurologic phenotypes: AÂgenotypeâ€“phenotype correlation study. Genetics in Medicine, 2023, 25, 37-48.	1.1	3
4491	Parallel functional annotation of cancer-associated missense mutations in histone methyltransferases. Scientific Reports, 2022, 12, .	1.6	2
4492	Targeted long-read sequencing allows for rapid identification of pathogenic disease-causing variants in retinoblastoma. Ophthalmic Genetics, 2022, 43, 762-770.	0.5	1
4493	Identification of a new familial case of 3q29 deletion syndrome associated with cleft lip and palate via wholeâ€exome sequencing. American Journal of Medical Genetics, Part A, 0, , .	0.7	0
4494	Expanding SPTAN1 monoallelic variant associated disorders: From epileptic encephalopathy to pure spastic paraplegia and ataxia. Genetics in Medicine, 2023, 25, 76-89.	1.1	3
4495	Concurrent Pathogenic Variants of BRCA1, MUTYH and CHEK2 in a Hereditary Cancer Family. Cancer Genetics, 2022, 268-269, 128-136.	0.2	1
4496	Inherited human ITK deficiency impairs IFN-Î³ immunity and underlies tuberculosis. Journal of Experimental Medicine, 2023, 220, .	4.2	8
4497	Applications of Long-Read Sequencing Technology in Clinical Genomics. Advances in Molecular Pathology, 2022, 5, 85-108.	0.2	0
4499	Systematic analysis and prediction of genes associated with monogenic disorders on human chromosome X. Nature Communications, 2022, 13, .	5.8	14
4500	A Human Hereditary Cardiomyopathy Shares a Genetic Substrate With Bicuspid Aortic Valve. Circulation, 2023, 147, 47-65.	1.6	9

#	Article	IF	Citations
4501	Characterization of Arabian Peninsula whole exomes: Contributing to the catalogue of human diversity. IScience, 2022, 25, 105336.	1.9	0
4502	A rare PRSS1 p.S127C mutation is associated with chronic pancreatitis and causes misfolding-induced ER-stress. Pancreatology, 2022, 22, 1112-1119.	0.5	3
4503	A missense variant in NCF1 is associated with susceptibility to unexplained recurrent spontaneous abortion. Open Life Sciences, 2022, 17, 1443-1450.	0.6	0
4504	Exploring the genetic landscape of diphtheria, tetanus and pertussis vaccination-associated seizures or subsequent epilepsies. , 2023, 8, 100094.		0
4505	IdentificaciÃ³n preliminar de variantes patÃ³genas en una familia raizal afrocolombiana con factores de riesgo para glaucoma. Colombia Medica, 2022, 53, e2065107.	0.7	0
4506	Counterintuitive structural and functional effects due to naturally occurring mutations targeting the active site of the diseaseâ€associated <scp>NQO1</scp> enzyme*. FEBS Journal, 2023, 290, 1855-1873.	2.2	1
4507	Predicting functional effect of missense variants using graph attention neural networks. Nature Machine Intelligence, 2022, 4, 1017-1028.	8.3	15
4508	Spectrum of DDC variants causing aromatic l-amino acid decarboxylase (AADC) deficiency and pathogenicity interpretation using ACMG-AMP/ACGS recommendations. Molecular Genetics and Metabolism, 2022, 137, 359-381.	0.5	9
4509	Partial loss-of-function mutations in GINS4 lead to NK cell deficiency with neutropenia. JCI Insight, 2022, 7, .	2.3	7
4510	Pathogenic Interleukin-10 Receptor Alpha Variants in Humans â€" Balancing Natural Selection and Clinical Implications. Journal of Clinical Immunology, 0, , .	2.0	1
4511	Deleterious Variation in Natural Populations and Implications for Conservation Genetics. Annual Review of Animal Biosciences, 2023, 11, 93-114.	3.6	32
4512	Assessment of an automated approach for variant interpretation in screening for monogenic disorders: A singleâ€center study. Molecular Genetics & Genomic Medicine, 0, , .	0.6	1
4513	Reconstructing mutational lineages in breast cancer by multi-patient-targeted single-cell DNA sequencing. Cell Genomics, 2023, 3, 100215.	3.0	4
4514	FAVOR: functional annotation of variants online resource and annotator for variation across the human genome. Nucleic Acids Research, 2023, 51, D1300-D1311.	6.5	39
4515	Genetic insights into smoking behaviours in 10,558 men of African ancestry from continental Africa and the UK. Scientific Reports, 2022, 12, .	1.6	1
4516	Clinical and Genetic Characterization of Patients with Artemis Deficiency in Japan. Journal of Clinical Immunology, 2023, 43, 585-594.	2.0	1
4518	Identification of PCSK9-like human gene knockouts using metabolomics, proteomics, and whole-genome sequencing in a consanguineous population. Cell Genomics, 2023, 3, 100218.	3.0	4
4519	Inherited cancer predisposing mutations in patients with therapyâ€related myeloid neoplasms. British Journal of Haematology, 2023, 200, 489-493.	1.2	1

#	Article	IF	Citations
4520	Deep learning-assisted genome-wide characterization of massively parallel reporter assays. Nucleic Acids Research, 0, , .	6.5	1
4521	Integrated gene analyses of de novo variants from 46,612 trios with autism and developmental disorders. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	3.3	18
4522	Genetic Diagnosis of Inborn Errors of Immunity in an Emerging Country: a Retrospective Study of 216ÂMoroccan Patients. Journal of Clinical Immunology, 2023, 43, 485-494.	2.0	5
4523	Carrier frequency and incidence estimation of familial hemophagocytic lymphohistiocytosis in East Asian populations by genome aggregation database (gnomAD) based analysis. Frontiers in Pediatrics, 0, 10, .	0.9	1
4524	Identification of genetic loci that overlap between schizophrenia and metabolic syndrome. Psychiatry Research, 2022, 318, 114947.	1.7	1
4525	Archaic introgression contributed to the pre-agriculture adaptation of vitamin B1 metabolism in East Asia. IScience, 2022, 25, 105614.	1.9	3
4526	Fetal skeletal dysplasia cohort of a single tertiary referral center in Istanbul, Turkey. American Journal of Medical Genetics, Part A, 0, , .	0.7	0
4527	Phenotypic screening models for rapid diagnosis of genetic variants and discovery of personalized therapeutics. Molecular Aspects of Medicine, 2023, 91, 101153.	2.7	7
4528	Prioritizing autoimmunity risk variants for functional analyses by fine-mapping mutations under natural selection. Nature Communications, 2022, 13, .	5.8	3
4529	Prioritization of non-coding elements involved in non-syndromic cleft lip with/without cleft palate through genome-wide analysis of de novo mutations. Human Genetics and Genomics Advances, 2023, 4, 100166.	1.0	2
4530	The performance of multi-gene panels for breast/ovarian cancer predisposition. Clinica Chimica Acta, 2023, 539, 151-161.	0.5	3
4531	A recessive form of craniodiaphyseal dysplasia caused by a homozygous missense variant in SP7/Osterix. Bone, 2023, 167, 116633.	1.4	1
4532	Reporting Two Novel Mutations in Two Iranian Families with Cystic Fibrosis, Molecular and Bioinformatic Analysis. Iranian Biomedical Journal, 2022, 26, 398-405.	0.4	1
4534	The role of TBX18 in congenital heart defects in humans not confirmed. European Journal of Human Genetics, 0, , .	1.4	0
4535	Congenital Stationary Night Blindness: Clinical and Genetic Features. International Journal of Molecular Sciences, 2022, 23, 14965.	1.8	4
4537	Structural deficits in key domains of Shank2 lead to alterations in postsynaptic nanoclusters and to a neurodevelopmental disorder in humans. Molecular Psychiatry, 0, , .	4.1	3
4538	Insights on variant analysis in silico tools for pathogenicity prediction. Frontiers in Genetics, 0, 13, .	1.1	17
4539	A KCNC1â€related neurological disorder due to gain of Kv3.1 function. Annals of Clinical and Translational Neurology, 2023, 10, 111-117.	1.7	3

#	Article	IF	Citations
4541	A Novel CDH1 Variant Identified in a Chinese Family with Blepharocheilodontic Syndrome. Diagnostics, 2022, 12, 2936.	1.3	2
4542	Biallelic Inactivating TUB Variants Cause Retinal Ciliopathy Impairing Biogenesis and the Structure of the Primary Cilium. International Journal of Molecular Sciences, 2022, 23, 14656.	1.8	0
4543	Genetic Variants in ARHGEF6 Cause Congenital Anomalies of the Kidneys and Urinary Tract in Humans, Mice, and Frogs. Journal of the American Society of Nephrology: JASN, 2023, 34, 273-290.	3.0	2
4544	Editorial: Towards genome interpretation: Computational methods to model the genotype-phenotype relationship. Frontiers in Bioinformatics, 0, 2, .	1.0	1
4545	Comprehensive Genetic Exploration of Fused Teeth by Whole Exome Sequencing. Applied Sciences (Switzerland), 2022, 12, 11899.	1.3	1
4546	Proteogenomic insights into the biology and treatment of pancreatic ductal adenocarcinoma. Journal of Hematology and Oncology, 2022, 15, .	6.9	9
4547	Pan-cancer illumination of TRIM gene family reveals immunology regulation and potential therapeutic implications. Human Genomics, 2022, 16, .	1.4	2
4548	Evaluation of in silico predictors on short nucleotide variants in HBA1, HBA2, and HBB associated with haemoglobinopathies. ELife, 0, 11, .	2.8	6
4550	Optimized high-throughput screening of non-coding variants identified from genome-wide association studies. Nucleic Acids Research, 2023, 51, e18-e18.	6.5	3
4551	Data-driven historical characterization of epilepsy-associated genes. European Journal of Paediatric Neurology, 2023, 42, 82-87.	0.7	11
4552	Tackling hypo and hyper sensory processing heterogeneity in autism: From clinical stratification to genetic pathways. Autism Research, 0, , .	2.1	4
4553	De novo CLCN3 variants affecting Gly327 cause severe neurodevelopmental syndrome with brain structural abnormalities. Journal of Human Genetics, 0, , .	1.1	1
4554	Disruption of the HIF-1 pathway in individuals with Ollier disease and Maffucci syndrome. PLoS Genetics, 2022, 18, e1010504.	1.5	2
4555	Exonic variants of the P2RX7 gene in familial multiple sclerosis. NeurologÃa (English Edition), 2022, , .	0.2	0
4556	The Genetic Spectrum of Maturity-Onset Diabetes of the Young (MODY) in Qatar, a Population-Based Study. International Journal of Molecular Sciences, 2023, 24, 130.	1.8	2
4557	Single-neuron whole genome sequencing identifies increased somatic mutation burden in Alzheimer's disease related genes. Neurobiology of Aging, 2023, 123, 222-232.	1.5	2
4558	Diverse monogenic subforms of human spermatogenic failure. Nature Communications, 2022, 13, .	5.8	17
4559	Targeted gene panel analysis of Japanese patients with maturityâ€onset diabetes of the youngâ€like diabetes mellitus: Roles of inactivating variants in the <scp>ABCC8</scp> and insulin resistance genes. Journal of Diabetes Investigation, 2023, 14, 387-403.	1.1	1

#	Article	IF	Citations
4560	A novel homozygous nonsense <scp> NDNF </scp> variant in Kallmann syndrome. American Journal of Medical Genetics, Part A, 0, , .	0.7	0
4561	SysBiolPGWAS: simplifying post-GWAS analysis through the use of computational technologies and integration of diverse omics datasets. Bioinformatics, 2023, 39, .	1.8	1
4562	KMT2Aâ€Ð pathogenicity, prevalence, and variation according to a population database. Cancer Medicine, 2023, 12, 7234-7245.	1.3	1
4563	Precision medicine: The use of tailored therapy in primary immunodeficiencies. Frontiers in Immunology, 0, 13, .	2.2	6
4564	Powerful, scalable and resource-efficient meta-analysis of rare variant associations in large whole genome sequencing studies. Nature Genetics, 2023, 55, 154-164.	9.4	12
4565	Optical genome mapping and revisiting short-read genome sequencing data reveal previously overlooked structural variants disrupting retinal diseaseâ˜associated genes. Genetics in Medicine, 2023, 25, 100345.	1.1	9
4566	Genome-wide association study for vascular aging highlights pathways shared with cardiovascular traits in Koreans. Frontiers in Cardiovascular Medicine, 0, 9, .	1.1	0
4567	Evaluating gap junction variants for a role in pediatric cataract: an overview of the genetic landscape and clinical classification of variants in the GJA3 and GJA8 genes. Expert Review of Ophthalmology, 2023, 18, 71-95.	0.3	1
4569	Measurement of FGFR3 signaling at the cell membrane via total internal reflection fluorescence microscopy to compare the activation of FGFR3 mutants. Journal of Biological Chemistry, 2023, 299, 102832.	1.6	2
4571	Pathogenic Variant Spectrum in Breast Cancer Risk Genes in Finnish Patients. Cancers, 2022, 14, 6158.	1.7	2
4572	A Novel ARMC5 Germline Variant in Primary Macronodular Adrenal Hyperplasia Using Whole-Exome Sequencing. Diagnostics, 2022, 12, 3028.	1.3	1
4573	Pathogenic/likely pathogenic copy number variations and regions of homozygosity in fetal central nervous system malformations. Archives of Gynecology and Obstetrics, 0, , .	0.8	1
4574	The collective effects of genetic variants and complex traits. Journal of Human Genetics, 2023, 68, 255-262.	1.1	2
4575	Whole exome sequencing of 28 families of Danish descent reveals novel candidate genes and pathways in developmental dysplasia of the hip. Molecular Genetics and Genomics, 0, , .	1.0	1
4576	Novel PGM3 mutation in two siblings with combined immunodeficiency and childhood bullous pemphigoid: a case report and review of the literature. Allergy, Asthma and Clinical Immunology, 2022, 18, .	0.9	2
4579	Biallelic variants in OGDH encoding oxoglutarate dehydrogenase lead to a neurodevelopmental disorder characterized by global developmental delay, movement disorder, and metabolic abnormalities. Genetics in Medicine, 2023, 25, 100332.	1.1	4
4581	A generalizable deep learning framework for inferring fine-scale germline mutation rate maps. Nature Machine Intelligence, 2022, 4, 1209-1223.	8.3	5
4582	De novo mutation hotspots in homologous protein domains identify function-altering mutations in neurodevelopmental disorders. American Journal of Human Genetics, 2023, 110, 92-104.	2.6	3

#	Article	IF	Citations
4583	Calibration of computational tools for missense variant pathogenicity classification and ClinGen recommendations for PP3/BP4 criteria. American Journal of Human Genetics, 2022, 109, 2163-2177.	2.6	124
4584	VPatho: a deep learning-based two-stage approach for accurate prediction of gain-of-function and loss-of-function variants. Briefings in Bioinformatics, 2023, 24, .	3.2	5
4585	Loss of the Immunomodulatory Transcription Factor BATF2 in Humans Is Associated with a Neurological Phenotype. Cells, 2023, 12, 227.	1.8	2
4586	Heterozygous mutations in SOX2 may cause idiopathic hypogonadotropic hypogonadism via dominant-negative mechanisms. JCI Insight, 2023, 8, .	2.3	0
4587	Whole-exome sequencing identifies FANC heterozygous germline mutation as an adverse factor for immunosuppressive therapy in Chinese aplastic anemia patients aged 40 or younger: a single-center retrospective study. Annals of Hematology, 0, , .	0.8	0
4588	Dr.Nod: computational framework for discovery of regulatory non-coding drivers in tissue-matched distal regulatory elements. Nucleic Acids Research, 2023, 51, e23-e23.	6.5	1
4589	Recent Advances in Genetic Epidemiology of Colorectal Cancer in Chinese Population. , 2022, , 187-214.		0
4590	Effective smMIPs-Based Sequencing of Maculopathy-Associated Genes in Stargardt Disease Cases and Allied Maculopathies from the UK. Genes, 2023, 14, 191.	1.0	1
4591	Novel candidate genes for cholesteatoma in chronic otitis media. Frontiers in Genetics, 0, 13, .	1.1	0
4593	CRISPR-Cas9 base editors and their current role in human therapeutics. Cytotherapy, 2023, 25, 270-276.	0.3	4
4594	Systemic structural analysis of alterations reveals a common structural basis of driver mutations in cancer. NAR Cancer, 2023, 5, .	1.6	1
4595	A novel NONO variant that causes developmental delay and cardiac phenotypes. Scientific Reports, 2023, 13, .	1.6	1
4597	Molecular Landscape of Touretteâ€™s Disorder. International Journal of Molecular Sciences, 2023, 24, 1428.	1.8	0
4598	Genetic Variant in GRM1 Underlies Congenital Cerebellar Ataxia with No Obvious Intellectual Disability. International Journal of Molecular Sciences, 2023, 24, 1551.	1.8	3
4599	PDGFRB and NOTCH3 Mutations are Detectable in a Wider Range of Pericytic Tumors, Including Myopericytomas, Angioleiomyomas, Glomus Tumors, and Their Combined Tumors. Modern Pathology, 2023, 36, 100070.	2.9	4
4601	Are Alzheimer's and coronary artery diseases genetically related to longevity?. Frontiers in Psychiatry, 0, 13, .	1.3	1
4602	Genome-Wide Association and Inheritance-Based Analyses Implicate Unconventional Myosin Genes in Hypoplastic Left Heart Syndrome. Circulation Genomic and Precision Medicine, 0, , .	1.6	0
4603	TRIM25 mutation (p.C168*), coding for an E3 ubiquitin ligase, is a cause of earlyâ€onset autosomal dominant dementia with amyloid load and parkinsonism. Alzheimer's and Dementia, 2023, 19, 2805-2815.	0.4	4

#	Article	IF	Citations
4604	Genetic overlap between Parkinsonâ€™s disease and inflammatory bowel disease. Brain Communications, 2022, 5, .	1.5	3
4605	Deleterious, protein-altering variants in the transcriptional coregulator ZMYM3 in 27 individuals with a neurodevelopmental delay phenotype. American Journal of Human Genetics, 2023, 110, 215-227.	2.6	4
4606	Predicting the functional effects of human non-coding variants based on stacking ensemble learning. , 2022, , .		0
4607	High-frequency and functional mitochondrial DNA mutations at the single-cell level. Proceedings of the National Academy of Sciences of the United States of America, 2023, 120, .	3.3	9
4608	378. Investigation of GWAS variants associated with loin depth in commercial pigs. , 2022, , .		0
4609	A genome-wide association study of a rage-related misophonia symptom and the genetic link with audiological traits, psychiatric disorders, and personality. Frontiers in Neuroscience, 0, 16, .	1.4	2
4610	Proteogenomics of diffuse gliomas reveal molecular subtypes associated with specific therapeutic targets and immune-evasion mechanisms. Nature Communications, 2023, 14, .	5.8	7
4611	Meta-Analysis and Multivariate GWAS Analyses in 80,950 Individuals of African Ancestry Identify Novel Variants Associated with Blood Pressure Traits. International Journal of Molecular Sciences, 2023, 24, 2164.	1.8	0
4612	Genome-Wide Association Study on Longitudinal Change in Fasting Plasma Glucose in Korean Population. Diabetes and Metabolism Journal, 0, , .	1.8	1
4614	Haploinsufficiency as a Foreground Pathomechanism of Poirer-Bienvenu Syndrome and Novel Insights Underlying the Phenotypic Continuum of CSNK2B-Associated Disorders. Genes, 2023, 14, 250.	1.0	2
4616	A Spontaneous Nonhuman Primate Model of Myopic Foveoschisis. , 2023, 64, 18.		3
4618	FANCM missense variants and breast cancer risk: a case-control association study of 75,156 European women. European Journal of Human Genetics, 2023, 31, 578-587.	1.4	2
4619	TIVAN-indel: a computational framework for annotating and predicting non-coding regulatory small insertions and deletions. Bioinformatics, 2023, 39, .	1.8	2
4620	Predicting mutational function using machine learning. Mutation Research - Reviews in Mutation Research, 2023, 791, 108457.	2.4	0
4621	The PER3rs772027021 SNP induces pigmentation phenotypes of dyschromatosis universalis hereditaria. Journal of Molecular Medicine, 2023, 101, 279-294.	1.7	2
4622	Next-generation sequencing of 500 POI patients identified novel responsible monogenic and oligogenic variants. Journal of Ovarian Research, 2023, 16, .	1.3	4
4623	<scp> PAK1 </scp> c. <scp>1409â€‰T</scp> Â>â€‰a (p. <scp>Leu470Gln</scp>) de novo variant affects the protein kinase domain, leading to epilepsy, macrocephaly, spastic quadriplegia, and hydrocephalus: Case report and review of the literature. American Journal of Medical Genetics, Part A, 0, , .	0.7	0
4624	The Genetic Architectures of Functional and Structural Connectivity Properties within Cerebral Resting-State Networks. ENeuro, 2023, 10, ENEURO.0242-22.2023.	0.9	2

#	Article	IF	Citations
4625	An Integrated Pipeline for Trioâ€Rapid Genome Sequencing in Critically Ill Infants. Current Protocols, 2023, 3, .	1.3	0
4626	Relevance of Coding Variation in FLG And DOCK8 in Finnish Pediatric Patients with Early-Onset Moderate-To-Severe Atopic Dermatitis. JID Innovations, 2023, 3, 100203.	1.2	1
4627	A heterozygous missense variant in DLX3 leads to uterine leiomyomas and pregnancy losses in a consanguineous Iranian family. Gene, 2023, 865, 147292.	1.0	0
4628	Polygenic influences on the behavioral effects of alcohol withdrawal in a mixed-ancestry population from the collaborative study on the genetics of alcoholism (COGA). Molecular and Cellular Neurosciences, 2023, 125, 103851.	1.0	1
4630	Whole-exome sequencing and electrophysiological study reveal a novel loss-of-function mutation of KCNA10 in epinephrine provoked long QT syndrome with familial history of sudden cardiac death. Legal Medicine, 2023, 62, 102245.	0.6	0
4631	Loci for insulin processing and secretion provide insight into type 2 diabetes risk. American Journal of Human Genetics, 2023, 110, 284-299.	2.6	9
4632	Using Cluster Analysis to Overcome the Limits of Traditional Phenotypeâ€“Genotype Correlations: The Example of RYR1-Related Myopathies. Genes, 2023, 14, 298.	1.0	5
4635	Decoding the genetic and epigenetic basis of asthma. Allergy: European Journal of Allergy and Clinical Immunology, 2023, 78, 940-956.	2.7	17
4636	Cost-effective sequence analysis of 113 genes in 1,192 probands with retinitis pigmentosa and Leber congenital amaurosis. Frontiers in Cell and Developmental Biology, 0, 11, .	1.8	4
4637	Targeted next-generation sequencing of genes involved in Warfarin Pharmacodynamics and pharmacokinetics pathways using the Saudi Warfarin Pharmacogenetic study (SWAP). Pharmacogenomics Journal, 0, , .	0.9	0
4638	Case report: Novel compound heterozygosity for pathogenic variants in MED23 in a syndromic patient with postnatal microcephaly. Frontiers in Neurology, 0, 14, .	1.1	1
4639	Growth patterns and outcomes of growth hormone therapy in patients with acrodysostosis. Journal of Endocrinological Investigation, 0, , .	1.8	1
4640	The â€œextreme phenotype approachâ€•applied to male breast cancer allows the identification of rare variants of ATR as potential breast cancer susceptibility alleles. Oncotarget, 2023, 14, 111-125.	0.8	0
4641	NMNAT1 and hereditary spastic paraplegia (HSP): expanding the phenotypic spectrum of NMNAT1 variants. Neuromuscular Disorders, 2023, 33, 295-301.	0.3	3
4642	Aberrant phase separation and nucleolar dysfunction in rare genetic diseases. Nature, 0, , .	13.7	9
4643	The Genotype-Phenotype Correlation in Human 5Î±-Reductase Type 2 Deficiency: Classified and Analyzed from a SRD5A2 Structural Perspective. International Journal of Molecular Sciences, 2023, 24, 3297.	1.8	2
4645	An ex vivo test to investigate genetic factors conferring susceptibility to atypical haemolytic uremic syndrome. Frontiers in Immunology, 0, 14, .	2.2	3
4646	Identification of novel genomic risk loci shared between common epilepsies and psychiatric disorders. Brain, 2023, 146, 3392-3403.	3.7	10

#	Article	IF	Citations
4647	Novel Pathogenic Mutations Identified from Whole-Genome Sequencing in Unsolved Cases of Patients Affected with Inherited Retinal Diseases. Genes, 2023, 14, 447.	1.0	4
4648	Human IL-23 is essential for IFN-Î³â€“dependent immunity to mycobacteria. Science Immunology, 2023, 8, .	5.6	28
4649	Systematic Assessment of Protein C-Termini Mutated in Human Disorders. Biomolecules, 2023, 13, 355.	1.8	0
4651	Functional Characterization of Six SLCO1B1 (OATP1B1) Variants Observed in Finnish Individuals with a Psychotic Disorder. Molecular Pharmaceutics, 2023, 20, 1500-1508.	2.3	1
4654	Whole Exome Sequencing Reveals Novel Candidate Genes in Familial Forms of Glaucomatous Neurodegeneration. Genes, 2023, 14, 495.	1.0	0
4655	Molecular Dynamic Simulation Analysis of a Novel Missense Variant in CYB5R3 Gene in Patients with Methemoglobinemia. Medicina (Lithuania), 2023, 59, 379.	0.8	5
4656	Variants in DTNA cause a mild, dominantly inherited muscular dystrophy. Acta Neuropathologica, 2023, 145, 479-496.	3.9	0
4657	Whole Exome Sequence Analysis for Inborn Errors of IL-12/IFN-Î³ Axis in Patient with Recurrent Typhoid Fever. BioMed Research International, 2023, 2023, 1-6.	0.9	1
4658	Germline NUP98 Variants in Two Siblings with a Rothmundâ€“Thomson-Like Spectrum: Protein Functional Changes Predicted by Molecular Modeling. International Journal of Molecular Sciences, 2023, 24, 4028.	1.8	2
4659	Proteome-wide Mendelian randomization implicates nephronectin as an actionable mediator of the effect of obesity on COVID-19 severity. Nature Metabolism, 2023, 5, 248-264.	5.1	24
4660	A Comprehensive Genetic Analysis of Slovenian Families with Multiple Cases of Orofacial Clefts Reveals Novel Variants in the Genes IRF6, GRHL3, and TBX22. International Journal of Molecular Sciences, 2023, 24, 4262.	1.8	0
4662	Identification of novel compound heterozygous variants in the SLC30A7 (ZNT7) gene in two French brothers with stunted growth, testicular hypoplasia and bone marrow failure. Human Molecular Genetics, 2023, 32, 2016-2031.	1.4	2
4663	Hacking hematopoiesis â€“ emerging tools for examining variant effects. DMM Disease Models and Mechanisms, 2023, 16, .	1.2	0
4664	Homozygous DBF4 mutation as a cause of severe congenital neutropenia. Journal of Allergy and Clinical Immunology, 2023, 152, 266-277.	1.5	2
4665	Multitrait genome-wide analyses identify new susceptibility loci and candidate drugs to primary sclerosing cholangitis. Nature Communications, 2023, 14, .	5.8	10
4666	Two new cases with novel pathogenic variants reflecting the clinical diversity of <scp>Schaafâ€Yang</scp> syndrome. Clinical Genetics, 2023, 104, 127-132.	1.0	0
4667	Unified views on variant impact across many diseases. Trends in Genetics, 2023, 39, 442-450.	2.9	3
4668	Expanding the phenome and variome of the ROBO-SLIT pathway in congenital heart defects: toward improving the genetic testing yield of CHD. Journal of Translational Medicine, 2023, 21, .	1.8	0

#	Article	IF	Citations
4670	Congenital hydrocephalus: new Mendelian mutations and evidence for oligogenic inheritance. Human Genomics, 2023, 17, .	1.4	4
4671	A clustering linear combination method for multiple phenotype association studies based on GWAS summary statistics. Scientific Reports, 2023, 13, .	1.6	0
4672	Evaluation of in silico pathogenicity prediction tools for the classification of small in-frame indels. BMC Medical Genomics, 2023, 16, .	0.7	1
4673	Whole exome sequencing in unexplained recurrent miscarriage families identified novel pathogenic genetic causes of euploid miscarriage. Human Reproduction, 2023, 38, 1003-1018.	0.4	3
4674	Minimal residual disease detection by mutation-specific droplet digital PCR for leukemia/lymphoma. International Journal of Hematology, 2023, 117, 910-918.	0.7	2
4675	Structural analysis of the P132L disease mutation in caveolin-1 reveals its role in the assembly of oligomeric complexes. Journal of Biological Chemistry, 2023, 299, 104574.	1.6	3
4676	Skewed X-chromosome inactivation in unsolved neurodevelopmental disease cases can guide re-evaluation For X-linked genes. European Journal of Human Genetics, 2023, 31, 1228-1236.	1.4	5
4677	Genome-Wide Associations and Confirmatory Meta-Analyses in Diabetic Retinopathy. Genes, 2023, 14, 653.	1.0	5
4678	Cytotoxic CD161â^'CD8+ TEMRA cells contribute to the pathogenesis of systemic lupus erythematosus. EBioMedicine, 2023, 90, 104507.	2.7	7
4681	The genetic architecture of human amygdala volumes and their overlap with common brain disorders. Translational Psychiatry, 2023, 13, .	2.4	2
4683	Shared genetic architecture between attention-deficit/hyperactivity disorder and lifespan. Neuropsychopharmacology, 2023, 48, 981-990.	2.8	1
4684	Genetic architecture of spatial electrical biomarkers for cardiac arrhythmia and relationship with cardiovascular disease. Nature Communications, 2023, 14, .	5.8	1
4686	Compound Heterozygous PLD1 Variants in Right-Sided Heart Malformations. Congenital Heart Disease, 2023, 18, 213-218.	0.0	0
4687	GWAS meta-analyses clarify the genetics of cervical phenotypes and inform risk stratification for cervical cancer. Human Molecular Genetics, 2023, 32, 2103-2116.	1.4	5
4688	Exomeâ€wide assessment of isolated biliary atresia: A report from the <scp>National Birth Defects Prevention Study</scp> using childâ€“parent trios and a caseâ€“control design to identify novel rare variants. American Journal of Medical Genetics, Part A, 2023, 191, 1546-1556.	0.7	2
4689	Genetic associations with age at dementia onset in the PSEN1 E280A Colombian kindred. Alzheimer's and Dementia, 2023, 19, 3835-3847.	0.4	2
4690	Molecular Landscape of Pelvic Organ Prolapse Provides Insights into Disease Etiology. International Journal of Molecular Sciences, 2023, 24, 6087.	1.8	4
4691	ADAM10 Gene Variants in AD Patients and Their Relationship to CSF Protein Levels. International Journal of Molecular Sciences, 2023, 24, 6113.	1.8	0

#	Article	IF	Citations
4692	Multiancestry genomic and transcriptomic analysis of gastric cancer. Nature Genetics, 2023, 55, 581-594.	9.4	6
4695	A thermosensitive PCNA allele underlies an ataxia-telangiectasia-like disorder. Journal of Biological Chemistry, 2023, 299, 104656.	1.6	4
4696	PredDSMC: A predictor for driver synonymous mutations in human cancers. Frontiers in Genetics, 0, 14, .	1.1	0
4697	Formin-mediated nuclear actin at androgen receptors promotes transcription. Nature, 2023, 617, 616-622.	13.7	9
4700	The Expanding Phenotypical Spectrum of WARS2-Related Disorder: Four Novel Cases with a Common Recurrent Variant. Genes, 2023, 14, 822.	1.0	0
4703	Autoimmunity and immunodeficiency associated with monoallelic LIG4 mutations via haploinsufficiency. Journal of Allergy and Clinical Immunology, 2023, , .	1.5	0
4704	Quantitative determination of SLC2A1 variant functional effects in GLUT1 deficiency syndrome. Annals of Clinical and Translational Neurology, 2023, 10, 787-801.	1.7	0
4705	Clinical Genetic Features and Neoadjuvant Chemotherapy Response in HER2-Low Breast Cancers: A Retrospective, Multicenter Cohort Study. Annals of Surgical Oncology, 2023, 30, 5653-5662.	0.7	4
4706	A novel compound heterozygous mutation of COL6A3 in Chinese patients with isolated cervical dystonia. Frontiers in Neurology, 0, 14, .	1.1	0
4707	Clinical and genetic analysis further delineates the phenotypic spectrum of ALDH1A3-related anophthalmia and microphthalmia. European Journal of Human Genetics, 0, , .	1.4	1
4709	The regulatory genome and defects in monogenic diseases. , 2023, , 275-302.		0
4711	Shared genetic loci and causal relations between schizophrenia and obsessive-compulsive disorder. , 2023, 9, .		1
4712	Identification of Rare Variants Involved in High Myopia Unraveled by Whole Genome Sequencing. Ophthalmology Science, 2023, 3, 100303.	1.0	0
4713	Dyslipidemia, inflammation, calcification, and adiposity in aortic stenosis: a genome-wide study. European Heart Journal, 2023, 44, 1927-1939.	1.0	19
4714	Novel ITGB6 variants cause hypoplastic-hypomineralized amelogenesis imperfecta and taurodontism: characterization of tooth phenotype and review of literature. BDJ Open, 2023, 9, .	0.8	3
4715	PredinID: Predicting Pathogenic Inframe Indels in Human Through Graph Convolution Neural Network With Graph Sampling Technique. IEEE/ACM Transactions on Computational Biology and Bioinformatics, 2023, 20, 3226-3233.	1.9	0
4716	A Novel Variant in VPS13B Underlying Cohen Syndrome. BioMed Research International, 2023, 2023, 1-7.	0.9	0
4717	Bi-allelic variants in INTS11 are associated with a complex neurological disorder. American Journal of Human Genetics, 2023, 110, 774-789.	2.6	8

#	Article	IF	Citations
4718	The origins and functional effects of postzygotic mutations throughout the human life span. Science, 2023, 380, .	6.0	6
4719	DrOGA: an artificial intelligence solution for driver-status prediction of genomics mutations in precision cancer medicine. IEEE Access, 2023, , 1-1.	2.6	0
4720	Genetic variants associated with spontaneous preterm birth in women from India: a prospective cohort study. , 2023, 14, 100190.		1
4721	The sequence kernel association test for multicategorical outcomes. Genetic Epidemiology, 2023, 47, 432-449.	0.6	0
4722	Combining globally search for a regular expression and print matching lines with bibliographic monitoring of genomic database improves diagnosis. Frontiers in Genetics, 0, 14, .	1.1	0
4723	A Comprehensive Analysis of 21 Actionable Pharmacogenes in the Spanish Population: From Genetic Characterisation to Clinical Impact. Pharmaceutics, 2023, 15, 1286.	2.0	2
4724	Prevalence of Monogenic Bone Disorders in a Dutch Cohort of Atypical Femur Fracture Patients. Journal of Bone and Mineral Research, 2020, 38, 896-906.	3.1	1
4725	Predicting the pathogenicity of missense variants using features derived from AlphaFold2. Bioinformatics, 2023, 39, .	1.8	13
4726	Systematic elucidation of genetic mechanisms underlying cholesterol uptake. Cell Genomics, 2023, , 100304.	3.0	2
4727	Next-generation sequencing for gene panels, clinical exome, and whole-genome analysis. , 2023, , 743-766.		0
4728	Contribution of LRP1 in Human Congenital Heart Disease Correlates with Its Roles in the Outflow Tract and Atrioventricular Cushion Development. Genes, 2023, 14, 947.	1.0	0
4729	eyeVarP: A computational framework for the identification of pathogenic variants specific to eyeÂdisease. Genetics in Medicine, 2023, 25, 100862.	1.1	2
4730	<scp>WWC2</scp> expression in the testis: Implications for spermatogenesis and male fertility. FASEB Journal, 2023, 37, .	0.2	0
4731	Encephalitis and poor neuronal deathâ€“mediated control of herpes simplex virus in human inherited RIPK3 deficiency. Science Immunology, 2023, 8, .	5.6	8
4732	POLG2-Linked Mitochondrial Disease: Functional Insights from New Mutation Carriers and Review of the Literature. Cerebellum, 2024, 23, 479-488.	1.4	1
4734	Whole-exome sequencing in moyamoya patients of Northern-European origin identifies gene variants involved in Nitric Oxide metabolism: A pilot study. Brain and Spine, 2023, 3, 101745.	0.0	1
4808	Artificial Intelligence and Its Integration with Regenerative Medicine Approach. Advances in Healthcare Information Systems and Administration Book Series, 2023, , 32-57.	0.2	1
4839	Case report: Expanding the phenotype of FOXP1-related intellectual disability syndrome and hyperkinetic movement disorder in differential diagnosis with epileptic seizures. Frontiers in Neurology, 0, 14, .	1.1	1

#	Article	IF	Citations
4901	Familial co-segregation and the emerging role of long-read sequencing to re-classify variants of uncertain significance in inherited retinal diseases. Npj Genomic Medicine, 2023, 8, .	1.7	1
4920	Case Report: Sequential postzygotic HRAS mutation and gains of the paternal chromosome 11 carrying the mutated allele in a patient with epidermal nevus and rhabdomyosarcoma: evidence of a multiple-hit mechanism involving HRAS in oncogenic transformation. Frontiers in Genetics, 0, 14, .	1.1	0
4939	Introduction to Machine Learning in Medicine. Imaging Informatics for Healthcare Professionals, 2023, , 39-68.	0.4	0
4940	Leber congenital amaurosis as the initial and essential manifestation in a Chinese patient with autoimmune polyglandular syndrome Type 1. Documenta Ophthalmologica, 0, , .	1.0	0
4951	Clinical and genetic analyses of a Swedish patient series diagnosed with ataxia. Journal of Neurology, 2024, 271, 526-542.	1.8	0
4979	Molecular complexity of diffuse large B-cell lymphoma: a molecular perspective and therapeutic implications. Journal of Applied Genetics, 0, , .	1.0	0
5037	Computational approaches for identifying disease-causing mutations in proteins. Advances in Protein Chemistry and Structural Biology, 2024, , 141-171.	1.0	0
5045	Next-generation sequencing and bioinformatics in rare movement disorders. Nature Reviews Neurology, 2024, 20, 114-126.	4.9	0
5088	Genetic and Genomic Results and Management. , 2024, , 93-110.		0
5112	Revolutionizing Genomics: Exploring the Potential of Next-Generation Sequencing. , 2024, , 1-33.		0