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Nucleic Acids Research

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Citation Report

#	ARTICLE	IF	CITATIONS
1	Annotating DNA Variants Is the Next Major Goal for Human Genetics. American Journal of Human Genetics, 2014, 94, 5-10.	6.2	32
2	Clinical Research Informatics for Big Data and Precision Medicine. Yearbook of Medical Informatics, 2016, 25, 211-218.	1.0	24
3	SAAMBE: Webserver to Predict the Charge of Binding Free Energy Caused by Amino Acids Mutations. International Journal of Molecular Sciences, 2016, 17, 547.	4.1	59
4	Epigenetic Regulations of GABAergic Neurotransmission: Relevance for Neurological Disorders and Epigenetic Therapy. Medical Epigenetics, 2016, 4, 1-19.	262.3	6,201
5	Preface - Access to Knowledge Revisited. Yearbook of Medical Informatics, 2016, 25, S18-S20.	1.0	1
6	Evaluation of Bioinformatic Programmes for the Analysis of Variants within Splice Site Consensus Regions. Advances in Bioinformatics, 2016, 2016, 1-10.	5.7	43
7	Assessing Photoreceptor Structure in Retinitis Pigmentosa and Usher Syndrome. , 2016, 57, 2428.		81
8	Challenges in Molecular Diagnostics of Channelopathies in the Next-Generation Sequencing Era: Less Is More?. Frontiers in Cardiovascular Medicine, 2016, 3, 29.	2.4	8
9	Colorectal Choriocarcinoma in a Patient with Probable Lynch Syndrome. Frontiers in Oncology, 2016, 6, 252.	2.8	5
10	Pathogenic Mutations in Cancer-Predisposing Genes: A Survey of 300 Patients with Whole-Genome Sequencing and Lifetime Electronic Health Records. PLoS ONE, 2016, 11, e0167847.	2.5	4
11	Whole Gene Capture Analysis of 15 CRC Susceptibility Genes in Suspected Lynch Syndrome Patients. PLoS ONE, 2016, 11, e0157381.	2.5	12
12	Type 2 diabetes: genetic data sharing to advance complex disease research. Nature Reviews Genetics, 2016, 17, 535-549.	16.3	128
13	The expanding phenotypic spectra of kidney diseases: insights from genetic studies. Nature Reviews Nephrology, 2016, 12, 472-483.	9.6	61
14	LSDBs and How They Have Evolved. Human Mutation, 2016, 37, 532-539.	2.5	6
15	Gene Variant Databases and Sharing: Creating a Global Genomic Variant Database for Personalized Medicine. Human Mutation, 2016, 37, 559-563.	2.5	24
16	OUP accepted manuscript. Nucleic Acids Research, 2017, 45, D626-D634.	14.5	308
17	Clinical implementation of genomic medicine: the importance of global collaboration. Expert Review of Precision Medicine and Drug Development, 2016, 1, 349-351.	0.7	3
18	Utilization of amplicon-based targeted sequencing panel for the massively parallel sequencing of sporadic hearing impairment patients from Saudi Arabia. BMC Medical Genetics, 2016, 17, 67.	2.1	11

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19	Identification and analysis of the human sex-biased genes. Briefings in Bioinformatics, 2018, 19, bbw125.	6.5	19
20	Response: Table 1.. Journal of the National Cancer Institute, 2016, 108, djw173.	6.3	2
21	Rare disruptive mutations in ciliary function genes contribute to testicular cancer susceptibility. Nature Communications, 2016, 7, 13840.	12.8	32
22	Exploring human disease using the Rat Genome Database. DMM Disease Models and Mechanisms, 2016, 9, 1089-1095.	2.4	27
23	Opening Pandora's Box – incidental genetic findings. Nature Reviews Cardiology, 2016, 13, 187-188.	13.7	0
24	Programmable editing of a target base in genomic DNA without double-stranded DNA cleavage. Nature, 2016, 533, 420-424.	27.8	3,662
25	Precision medicine for psychopharmacology: a general introduction. Expert Review of Neurotherapeutics, 2016, 16, 831-839.	2.8	15
26	G23D: Online tool for mapping and visualization of genomic variants on 3D protein structures. BMC Genomics, 2016, 17, 681.	2.8	18
27	Inherited platelet disorders: toward DNA-based diagnosis. Blood, 2016, 127, 2814-2823.	1.4	119
28	A Novel Mutation in a Critical Region for the Methyl Donor Binding in DNMT3B Causes Immunodeficiency, Centromeric Instability, and Facial Anomalies Syndrome (ICF). Journal of Clinical Immunology, 2016, 36, 801-809.	3.8	12
29	Medullary Thyroid Carcinoma Associated with Germline <i>RET</i> ^{K666N} Mutation. Thyroid, 2016, 26, 1744-1751.	4.5	7
30	Immunogenomics of Hypermutated Glioblastoma: A Patient with Germline <i>POLE</i> Deficiency Treated with Checkpoint Blockade Immunotherapy. Cancer Discovery, 2016, 6, 1230-1236.	9.4	242
31	Mutation analysis by direct and whole exome sequencing in familial and sporadic tooth agenesis. International Journal of Molecular Medicine, 2016, 38, 1338-1348.	4.0	22
32	User-centered design of multi-gene sequencing panel reports for clinicians. Journal of Biomedical Informatics, 2016, 63, 1-10.	4.3	18
33	Proteome-wide Structural Analysis of PTM Hotspots Reveals Regulatory Elements Predicted to Impact Biological Function and Disease. Molecular and Cellular Proteomics, 2016, 15, 3513-3528.	3.8	31
34	Conflicting Interpretation of Genetic Variants and Cancer Risk by Commercial Laboratories as Assessed by the Prospective Registry of Multiplex Testing. Journal of Clinical Oncology, 2016, 34, 4071-4078.	1.6	147
35	REVEL: An Ensemble Method for Predicting the Pathogenicity of Rare Missense Variants. American Journal of Human Genetics, 2016, 99, 877-885.	6.2	1,555
36	Retrieving GPCR data from public databases. Current Opinion in Pharmacology, 2016, 30, 38-43.	3.5	4

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37	Molecular Testing in Breast Cancer: A Guide to Current Practices. Archives of Pathology and Laboratory Medicine, 2016, 140, 815-824.	2.5	43
38	Implementing genomics and pharmacogenomics in the clinic: The National Human Genome Research Institute's genomic medicine portfolio. Atherosclerosis, 2016, 253, 225-236.	0.8	23
39	Homozygous inactivation of <i>CHEK2</i> is linked to a familial case of multiple primary lung cancer with accompanying cancers in other organs. Journal of Physical Education and Sports Management, 2016, 2, a001032.	1.2	16
40	The power of multiplexed functional analysis of genetic variants. Nature Protocols, 2016, 11, 1782-1787.	12.0	115
41	IMHOTEP—a composite score integrating popular tools for predicting the functional consequences of non-synonymous sequence variants. Nucleic Acids Research, 2017, 45, gkw886.	14.5	10
42	Genetic Elucidation of Nonsyndromic Hearing Loss in the High-Throughput Sequencing Era. Monographs in Human Genetics, 0, , 56-72.	0.5	3
43	Selective RNA targeting and regulated signaling by RIG-I is controlled by coordination of RNA and ATP binding. Nucleic Acids Research, 2016, 45, gkw816.	14.5	15
44	Actionable Genes, Core Databases, and Locus-Specific Databases. Human Mutation, 2016, 37, 1299-1307.	2.5	6
45	WES/WGS Reporting of Mutations from Cardiovascular “Actionable” Genes in Clinical Practice: A Key Role for UMD Knowledgebases in the Era of Big Databases. Human Mutation, 2016, 37, 1308-1317.	2.5	5
46	Analysis of large-scale whole exome sequencing data to determine the prevalence of genetically-distinct forms of neuronal ceroid lipofuscinosis. Gene, 2016, 593, 284-291.	2.2	31
47	3Disease Browser: A Web server for integrating 3D genome and disease-associated chromosome rearrangement data. Scientific Reports, 2016, 6, 34651.	3.3	32
48	The Allelic Landscape of Human Blood Cell Trait Variation and Links to Common Complex Disease. Cell, 2016, 167, 1415-1429.e19.	28.9	1,052
49	Somatic cancer variant curation and harmonization through consensus minimum variant level data. Genome Medicine, 2016, 8, 117.	8.2	61
50	Analysis of Arabidopsis Accessions Hypersensitive to a Loss of Chloroplast Translation. Plant Physiology, 2016, 172, 1862-1875.	4.8	21
51	Prevalence of low-penetrant germline TP53 D49H mutation in Japanese cancer patients. Biomedical Research, 2016, 37, 259-264.	0.9	8
52	Discrepancies between human DNA, mRNA and protein reference sequences and their relation to single nucleotide variants in the human population. Database: the Journal of Biological Databases and Curation, 2016, 2016, baw124.	3.0	4
53	StructMAN: annotation of single-nucleotide polymorphisms in the structural context. Nucleic Acids Research, 2016, 44, W463-W468.	14.5	32
54	Consideration of Cosegregation in the Pathogenicity Classification of Genomic Variants. American Journal of Human Genetics, 2016, 98, 1077-1081.	6.2	205

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55	A novel mutation in CELSR1 is associated with hereditary lymphedema. Vascular Cell, 2016, 8, 1.	0.2	38
56	Defining the Clinical Value of a Genomic Diagnosis in the Era of Next-Generation Sequencing. Annual Review of Genomics and Human Genetics, 2016, 17, 303-332.	6.2	43
57	From case studies to community knowledge base: MSeqDR provides a platform for the curation and genomic analysis of mitochondrial diseases. Journal of Physical Education and Sports Management, 2016, 2, a001065.	1.2	10
58	Targeted resequencing identifies <i>PTCH1</i> as a major contributor to ocular developmental anomalies and extends the SOX2 regulatory network. Genome Research, 2016, 26, 474-485.	5.5	37
59	ClinGen Pathogenicity Calculator: a configurable system for assessing pathogenicity of genetic variants. Genome Medicine, 2017, 9, 3.	8.2	59
60	MalaCards: an amalgamated human disease compendium with diverse clinical and genetic annotation and structured search. Nucleic Acids Research, 2017, 45, D877-D887.	14.5	398
61	CIViC is a community knowledgebase for expert crowdsourcing the clinical interpretation of variants in cancer. Nature Genetics, 2017, 49, 170-174.	21.4	460
62	Child&Parent Familial Hypercholesterolemia Screening in Primary Care. New England Journal of Medicine, 2017, 376, 498-500.	27.0	11
63	The current state of clinical interpretation of sequence variants. Current Opinion in Genetics and Development, 2017, 42, 33-39.	3.3	77
64	Whole-genome view of the consequences of a population bottleneck using 2926 genome sequences from Finland and United Kingdom. European Journal of Human Genetics, 2017, 25, 477-484.	2.8	60
65	DISCOVERY OF FUNCTIONAL AND DISEASE PATHWAYS BY COMMUNITY DETECTION IN PROTEIN-PROTEIN INTERACTION NETWORKS. , 2017, 22, 336-347.		7
66	Mutation effects predicted from sequence co-variation. Nature Biotechnology, 2017, 35, 128-135.	17.5	543
67	InterVar: Clinical Interpretation of Genetic Variants by the 2015 ACMG-AMP Guidelines. American Journal of Human Genetics, 2017, 100, 267-280.	6.2	717
68	GAVIN: Gene-Aware Variant Interpretation for medical sequencing. Genome Biology, 2017, 18, 6.	8.8	55
69	The Design of the Valsartan for Attenuating Disease Evolution in Early Sarcomeric Hypertrophic Cardiomyopathy (VANISH) Trial. American Heart Journal, 2017, 187, 145-155.	2.7	41
70	A de novo Pericentric Inversion in Chromosome 4 Associated with Disruption of PITX2 and a Microdeletion in 4p15.2 in a Patient with Axenfeld-Rieger Syndrome and Developmental Delay. Cytogenetic and Genome Research, 2017, 151, 5-9.	1.1	3
71	Quantitation of Targetable Somatic Mutations Among Patients Evaluated by a Personalized Medicine Clinical Service: Considerations for Off&Label Drug Use. Pharmacotherapy, 2017, 37, 1043-1051.	2.6	6
72	The Burden of Early Phenotypes and the Influence of Wall Thickness in Hypertrophic Cardiomyopathy Mutation Carriers. JAMA Cardiology, 2017, 2, 419.	6.1	50

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73	Genetic variants of the DNA repair genes from Exome Aggregation Consortium (EXAC) database: significance in cancer. <i>DNA Repair</i> , 2017, 52, 92-102.	2.8	13
74	Evaluating Variant Calling Tools for Non-Matched Next-Generation Sequencing Data. <i>Scientific Reports</i> , 2017, 7, 43169.	3.3	185
75	A Model Program for Translational Medicine in Epilepsy Genetics. <i>Journal of Child Neurology</i> , 2017, 32, 429-436.	1.4	6
76	Clinical Genetics Testing Laboratories Have a Remarkably Low Rate of Clinically Significant Discordance When Interpreting Variants in Hereditary Cancer Syndrome Genes. <i>Journal of Clinical Oncology</i> , 2017, 35, 1259-1261.	1.6	11
77	Rapid and cost-effective high-throughput sequencing for identification of germline mutations of BRCA1 and BRCA2. <i>Journal of Human Genetics</i> , 2017, 62, 561-567.	2.3	17
78	Whole Exome Sequencing Reveals Severe Thrombophilia in Acute Unprovoked Idiopathic Fatal Pulmonary Embolism. <i>EBioMedicine</i> , 2017, 17, 95-100.	6.1	13
79	Implementing Genome-Driven Oncology. <i>Cell</i> , 2017, 168, 584-599.	28.9	405
80	Germline mutations in patients with multiple colorectal polyps in China. <i>Journal of Gastroenterology and Hepatology (Australia)</i> , 2017, 32, 1723-1729.	2.8	6
81	LDLR Variant Databases and Familial Hypercholesterolemia Population Studies. <i>Journal of the American College of Cardiology</i> , 2017, 69, 754-755.	2.8	3
82	Increasing the genome-targeting scope and precision of base editing with engineered Cas9-cytidine deaminase fusions. <i>Nature Biotechnology</i> , 2017, 35, 371-376.	17.5	609
83	Opportunities and challenges of whole-genome and -exome sequencing. <i>BMC Genetics</i> , 2017, 18, 14.	2.7	160
84	Whole-genome landscape of pancreatic neuroendocrine tumours. <i>Nature</i> , 2017, 543, 65-71.	27.8	716
85	Links between critical proteins drive the controllability of protein interaction networks. <i>Proteomics</i> , 2017, 17, e1700056.	2.2	20
86	Research in Computational Molecular Biology. <i>Lecture Notes in Computer Science</i> , 2017, 10229, 389-390.	1.3	1
87	Whole-exome sequencing on deceased fetuses with ultrasound anomalies: expanding our knowledge of genetic disease during fetal development. <i>Genetics in Medicine</i> , 2017, 19, 1171-1178.	2.4	121
88	Quantifying the Impact of Non-coding Variants on Transcription Factor-DNA Binding. <i>Lecture Notes in Computer Science</i> , 2017, 10229, 336-352.	1.3	16
89	Clinical Variant Classification: A Comparison of Public Databases and a Commercial Testing Laboratory. <i>Oncologist</i> , 2017, 22, 797-803.	3.7	40
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91	The Proteins API: accessing key integrated protein and genome information. Nucleic Acids Research, 2017, 45, W539-W544.	14.5	69
92	Determination of disease phenotypes and pathogenic variants from exome sequence data in the CAGI 4 gene panel challenge. Human Mutation, 2017, 38, 1201-1216.	2.5	5
93	Melorheostosis: Exome sequencing of an associated dermatosis implicates postzygotic mosaicism of mutated KRAS. Bone, 2017, 101, 145-155.	2.9	37
94	Concordance between Research Sequencing and Clinical Pharmacogenetic Genotyping in the eMERGE-PCx Study. Journal of Molecular Diagnostics, 2017, 19, 561-566.	2.8	18
95	Application of Panel-Based Tests for Inherited Risk of Cancer. Annual Review of Genomics and Human Genetics, 2017, 18, 201-227.	6.2	26
96	Use of precision methods to accelerate drug development in oncology. Expert Review of Precision Medicine and Drug Development, 2017, 2, 109-120.	0.7	0
97	MARRVEL: Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome. American Journal of Human Genetics, 2017, 100, 843-853.	6.2	181
98	Comprehensive population-wide analysis of Lynch syndrome in Iceland reveals founder mutations in MSH6 and PMS2. Nature Communications, 2017, 8, 14755.	12.8	96
99	Assessment of the ExAC data set for the presence of individuals with pathogenic genotypes implicated in severe Mendelian pediatric disorders. Genetics in Medicine, 2017, 19, 1300-1308.	2.4	58
100	Clinical Genomic Testing. , 2017, , 247-262.		0
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102	Whole genome sequencing predicts novel human disease models in rhesus macaques. Genomics, 2017, 109, 214-220.	2.9	28
103	DATA SHARING AND REPRODUCIBLE CLINICAL GENETIC TESTING: SUCCESSES AND CHALLENGES. , 2017, 22, 166-176.		7
104	Utilization of genomic sequencing for population screening of immunodeficiencies in the newborn. Genetics in Medicine, 2017, 19, 1367-1375.	2.4	23
105	Exploring the global landscape of genetic variation in coagulation factor XI deficiency. Blood, 2017, 130, e1-e6.	1.4	41
106	Identification of a novel <i>RASD1</i> somatic mutation in a <i>USP8</i> -mutated corticotroph adenoma. Journal of Physical Education and Sports Management, 2017, 3, a001602.	1.2	8
107	PathOS: a decision support system for reporting high throughput sequencing of cancers in clinical diagnostic laboratories. Genome Medicine, 2017, 9, 38.	8.2	25
108	CAGI4 SickKids clinical genomes challenge: A pipeline for identifying pathogenic variants. Human Mutation, 2017, 38, 1169-1181.	2.5	11

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109	GenomeVIP: a cloud platform for genomic variant discovery and interpretation. <i>Genome Research</i> , 2017, 27, 1450-1459.	5.5	15
110	GeMSTONE: orchestrated prioritization of human germline mutations in the cloud. <i>Nucleic Acids Research</i> , 2017, 45, W207-W214.	14.5	2
111	Somatic Tumor Mutations Detected by Targeted Next Generation Sequencing in Minute Amounts of Serum-Derived Cell-Free DNA. <i>Scientific Reports</i> , 2017, 7, 2136.	3.3	7
112	A retrospective chart review of the features of PTEN hamartoma tumour syndrome in children. <i>Journal of Medical Genetics</i> , 2017, 54, 471-478.	3.2	87
113	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. <i>Nature Neuroscience</i> , 2017, 20, 1043-1051.	14.8	152
114	Deleterious variants in TRAK1 disrupt mitochondrial movement and cause fatal encephalopathy. <i>Brain</i> , 2017, 140, 568-581.	7.6	53
115	Ensemble variant interpretation methods to predict enzyme activity and assign pathogenicity in the CAG14 <i>NAGLU</i> (Human N-Acetylglucosaminidase) and <i>UBE2I</i> (Human SUMO1 ligase) challenges. <i>Human Mutation</i> , 2017, 38, 1109-1122.	2.5	14
116	Molecular diagnostics for hereditary hearing loss in children. <i>Expert Review of Molecular Diagnostics</i> , 2017, 17, 751-760.	3.1	21
117	Genetic Testing in the Evaluation of Unexplained Cardiac Arrest. <i>Circulation: Cardiovascular Genetics</i> , 2017, 10, .	5.1	76
118	Loss-of-function mutations in the CABLES1 gene are a novel cause of Cushing's disease. <i>Endocrine-Related Cancer</i> , 2017, 24, 379-392.	3.1	66
119	PMut: a web-based tool for the annotation of pathological variants on proteins, 2017 update. <i>Nucleic Acids Research</i> , 2017, 45, W222-W228.	14.5	184
120	A novel mutation in <i>GMPPA</i> in siblings with apparent intellectual disability, epilepsy, dysmorphism, and autonomic dysfunction. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2246-2250.	1.2	9
121	Sources of discordance among germ-line variant classifications in ClinVar. <i>Genetics in Medicine</i> , 2017, 19, 1118-1126.	2.4	88
122	Genomic diagnosis for children with intellectual disability and/or developmental delay. <i>Genome Medicine</i> , 2017, 9, 43.	8.2	188
123	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. <i>Bioinformatics</i> , 2017, 33, 1621-1629.	4.1	11
124	The association between germline <sc><i>BRCA2</i></sc> variants and sensitivity to platinum-based chemotherapy among men with metastatic prostate cancer. <i>Cancer</i> , 2017, 123, 3532-3539.	4.1	217
125	Accurately annotate compound effects of genetic variants using a context-sensitive framework. <i>Nucleic Acids Research</i> , 2017, 45, e82-e82.	14.5	9
126	Diagnosis of inherited bleeding disorders in the genomic era. <i>British Journal of Haematology</i> , 2017, 179, 363-376.	2.5	52

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127	Genetic predisposition to hematologic malignancies: management and surveillance. <i>Blood</i> , 2017, 130, 424-432.	1.4	145
128	Data resources for the identification and interpretation of actionable mutations by clinicians. <i>Annals of Oncology</i> , 2017, 28, 946-957.	1.2	20
129	Intragenic deletion of the <i>WDR45</i> gene in a male with encephalopathy, severe psychomotor disability, and epilepsy. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1444-1446.	1.2	10
130	Bedside Back to Bench: Building Bridges between Basic and Clinical Genomic Research. <i>Cell</i> , 2017, 169, 6-12.	28.9	103
131	Validation of copy number variation analysis for next-generation sequencing diagnostics. <i>European Journal of Human Genetics</i> , 2017, 25, 719-724.	2.8	72
132	Successful Application of Whole Genome Sequencing in a Medical Genetics Clinic. <i>Journal of Pediatric Genetics</i> , 2017, 06, 061-076.	0.7	54
133	Pathogenic variants in the healthy elderly: unique ethical and practical challenges. <i>Journal of Medical Ethics</i> , 2017, 43, 714-722.	1.8	10
134	The Human Gene Mutation Database: towards a comprehensive repository of inherited mutation data for medical research, genetic diagnosis and next-generation sequencing studies. <i>Human Genetics</i> , 2017, 136, 665-677.	3.8	1,106
135	Electronic Medical Record-Integrated Pharmacogenomics and Related Clinical Decision Support Concepts. <i>Clinical Pharmacology and Therapeutics</i> , 2017, 102, 254-264.	4.7	67
136	Development and Validation of Targeted Next-Generation Sequencing Panels for Detection of Germline Variants in Inherited Diseases. <i>Archives of Pathology and Laboratory Medicine</i> , 2017, 141, 787-797.	2.5	35
137	Clinical laboratories collaborate to resolve differences in variant interpretations submitted to ClinVar. <i>Genetics in Medicine</i> , 2017, 19, 1096-1104.	2.4	200
138	Genetics of Multiple Endocrine Neoplasia Type 1/Multiple Endocrine Neoplasia Type 2 Syndromes. <i>Endocrinology and Metabolism Clinics of North America</i> , 2017, 46, 491-502.	3.2	6
139	Further evidence that variants in <i>PPP1CB</i> cause a rasopathy similar to Noonan syndrome with loose anagen hair. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 565-567.	1.2	20
140	Germline Mutations in PALB2, BRCA1, and RAD51C, Which Regulate DNA Recombination Repair, in Patients With Gastric Cancer. <i>Gastroenterology</i> , 2017, 152, 983-986.e6.	1.3	98
141	Expanded national database collection and data coverage in the FINDbase worldwide database for clinically relevant genomic variation allele frequencies. <i>Nucleic Acids Research</i> , 2017, 45, D846-D853.	14.5	18
142	DisGeNET: a comprehensive platform integrating information on human disease-associated genes and variants. <i>Nucleic Acids Research</i> , 2017, 45, D833-D839.	14.5	1,865
143	The Monarch Initiative: an integrative data and analytic platform connecting phenotypes to genotypes across species. <i>Nucleic Acids Research</i> , 2017, 45, D712-D722.	14.5	306
144	The novel homozygous <i>KCNJ10</i> c.986T>C (p.(Leu329Pro)) variant is pathogenic for the SeSAME/EAST homologue in Malinois dogs. <i>European Journal of Human Genetics</i> , 2017, 25, 222-226.	2.8	16

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145	Programmable base editing of A→T to G→C in genomic DNA without DNA cleavage. <i>Nature</i> , 2017, 551, 464-471.	27.8	2,807
146	Identification of susceptible genes for complex chronic diseases based on disease risk functional SNPs and interaction networks. <i>Journal of Biomedical Informatics</i> , 2017, 74, 137-144.	4.3	6
147	Methods and Applications of CRISPR-Mediated Base Editing in Eukaryotic Genomes. <i>Molecular Cell</i> , 2017, 68, 26-43.	9.7	199
148	MAPPIN: a method for annotating, predicting pathogenicity and mode of inheritance for nonsynonymous variants. <i>Nucleic Acids Research</i> , 2017, 45, 10393-10402.	14.5	15
149	Copy Number Variation in Syndromic Forms of Psychiatric Illness: The Emerging Value of Clinical Genetic Testing in Psychiatry. <i>American Journal of Psychiatry</i> , 2017, 174, 1036-1050.	7.2	16
150	Pathogenicity of Hypertrophic Cardiomyopathy Variants. <i>Circulation: Cardiovascular Genetics</i> , 2017, 10, .	5.1	10
151	Repeat genetic testing with targeted capture sequencing in primary arrhythmia syndrome and cardiomyopathy. <i>European Journal of Human Genetics</i> , 2017, 25, 1313-1323.	2.8	9
152	The impact of rare variation on gene expression across tissues. <i>Nature</i> , 2017, 550, 239-243.	27.8	229
153	Genomic analysis of an infant with intractable diarrhea and dilated cardiomyopathy. <i>Journal of Physical Education and Sports Management</i> , 2017, 3, a002055.	1.2	13
154	Forces and Disease: Electrostatic force differences caused by mutations in kinesin motor domains can distinguish between disease-causing and non-disease-causing mutations. <i>Scientific Reports</i> , 2017, 7, 8237.	3.3	30
155	The Role of Hereditary Factors in Ovarian Carcinoma. <i>Clinical Obstetrics and Gynecology</i> , 2017, 60, 728-737.	1.1	1
156	Onco-proteogenomics: Multi-omics level data integration for accurate phenotype prediction. <i>Critical Reviews in Clinical Laboratory Sciences</i> , 2017, 54, 414-432.	6.1	16
157	PCSK9 inhibitor therapy in homozygous familial defective apolipoprotein B-100 due to APOB R3500Q: A case report. <i>Journal of Clinical Lipidology</i> , 2017, 11, 1471-1474.	1.5	4
158	Analysis and Annotation of Whole-Genome or Whole-Exome Sequencing Derived Variants for Clinical Diagnosis. <i>Current Protocols in Human Genetics</i> , 2017, 95, 9.24.1-9.24.28.	3.5	20
159	Characterisation of the novel deleterious RAD51C p.Arg312Trp variant and prioritisation criteria for functional analysis of RAD51C missense changes. <i>British Journal of Cancer</i> , 2017, 117, 1048-1062.	6.4	12
160	Inherited, not acquired, Gitelman syndrome in a patient with Sjögren's syndrome: importance of genetic testing to distinguish the two forms. <i>CEN Case Reports</i> , 2017, 6, 180-184.	0.9	12
161	The Use of Variant Maps to Explore Domain-Specific Mutations of FGFR1. <i>Journal of Dental Research</i> , 2017, 96, 1339-1345.	5.2	7
162	AgRP-LRP4-MuSK signaling as a therapeutic target for myasthenia gravis and other neuromuscular disorders. <i>Expert Opinion on Therapeutic Targets</i> , 2017, 21, 949-958.	3.4	44

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163	Frequency of mutations in a large series of clinically ascertained ovarian cancer cases tested on multi-gene panels compared to reference controls. <i>Gynecologic Oncology</i> , 2017, 147, 375-380.	1.4	105
164	Mutational profile of rare variants in inflammasome-related genes in Behçet disease: A Next Generation Sequencing approach. <i>Scientific Reports</i> , 2017, 7, 8453.	3.3	29
165	FOXP1 haploinsufficiency: Phenotypes beyond behavior and intellectual disability?. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 3172-3181.	1.2	18
166	Targeted sequencing of 36 known or putative colorectal cancer susceptibility genes. <i>Molecular Genetics & Genomic Medicine</i> , 2017, 5, 553-569.	1.2	32
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