

ClinVar: improving access to variant interpretations and

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Citation Report

#	ARTICLE	IF	CITATIONS
1	The 2018 Nucleic Acids Research database issue and the online molecular biology database collection. Nucleic Acids Research, 2018, 46, D1-D7.	6.5	106
2	The relationship between MnSOD Val16Ala gene polymorphism and the level of serum total antioxidant capacity with the risk of chronic kidney disease in type 2 diabetic patients: a nested case-control study in the Tehran lipid glucose study. Nutrition and Metabolism, 2018, 15, 25.	1.3	12
3	Psychological Impact of Learning & CDKN2A Variant Status as a Genetic Research Result. Public Health Genomics, 2018, 21, 154-163.	0.6	7
4	MAGUS: A Shared Tool for the Genetic Community. Circulation: Cardiovascular Quality and Outcomes, 2018, 11, e005006.	0.9	0
5	Rapid communication of efforts to resolve differences or update variant interpretations in ClinVar through case-level data sharing. Journal of Physical Education and Sports Management, 2018, 4, a003467.	0.5	2
6	TADeus-a tool for clinical interpretation of structural variants modifying chromatin organization. , 2018, , .		3
7	The value of genomic variant ClinVar submissions from clinical providers: Beyond the addition of novel variants. Human Mutation, 2018, 39, 1660-1667.	1.1	14
8	Dysplastic Lipoma. American Journal of Surgical Pathology, 2018, 42, 1530-1540.	2.1	36
9	Pleiotropic Phenotypes Associated With PKP2 Variants. Frontiers in Cardiovascular Medicine, 2018, 5, 184.	1.1	23
10	Comprehensive Study of the Clinical Phenotype of Germline <i>BAP1</i> Variant-Carrying Families Worldwide. Journal of the National Cancer Institute, 2018, 110, 1328-1341.	3.0	164
11	Progress in <i>BRCA</i> -Mutated Ovarian Cancer. New England Journal of Medicine, 2018, 379, 2567-2568.	13.9	18
12	Germline Variants in MLH1, MSH2, and MSH6 in Korean Patients with Lynch Syndrome. Laboratory Medicine Online, 2018, 8, 156.	0.0	2
13	A moderate form of osteogenesis imperfecta caused by compound heterozygous LEPRE1 mutations. Bone Reports, 2018, 9, 132-135.	0.2	7
14	Familial hypercholesterolemia in Canada: Initial results from the FH Canada national registry. Atherosclerosis, 2018, 277, 419-424.	0.4	18
15	Next-Generation Sequencing to Diagnose Suspected Genetic Disorders. New England Journal of Medicine, 2018, 379, 1353-1362.	13.9	181
16	ClinGen's GenomeConnect registry enables patient-centered data sharing. Human Mutation, 2018, 39, 1668-1676.	1.1	25
17	Variants in <i>NKX2-5</i> and <i>FLNC</i> Cause Dilated Cardiomyopathy and Sudden Cardiac Death. Circulation Genomic and Precision Medicine, 2018, 11, e002151.	1.6	27
18	Updated recommendation for the benign stand-alone ACMG/AMP criterion. Human Mutation, 2018, 39, 1525-1530.	1.1	102

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19	ClinVar at five years: Delivering on the promise. <i>Human Mutation</i> , 2018, 39, 1623-1630.	1.1	159
20	Genomic Data Management in Big Data Environments: The Colorectal Cancer Case. <i>Lecture Notes in Computer Science</i> , 2018, , 319-329.	1.0	3
21	Scaling resolution of variant classification differences in ClinVar between 41 clinical laboratories through an outlier approach. <i>Human Mutation</i> , 2018, 39, 1641-1649.	1.1	50
22	Bioinformatics in Clinical Genomic Sequencing. <i>Advances in Molecular Pathology</i> , 2018, 1, 9-26.	0.2	1
23	Prolonged Idasanutlin (RG7388) Treatment Leads to the Generation of p53-Mutated Cells. <i>Cancers</i> , 2018, 10, 396.	1.7	49
24	Copy Number Variants Account for a Tiny Fraction of Undiagnosed Myopathic Patients. <i>Genes</i> , 2018, 9, 524.	1.0	7
25	Web-Based Model for Predicting Time to Surgery in Young Patients with Familial Adenomatous Polyposis: An Internally Validated Study. <i>American Journal of Gastroenterology</i> , 2018, 113, 1881-1890.	0.2	8
26	Identification of Lynch syndrome risk variants in the Romanian population. <i>Journal of Cellular and Molecular Medicine</i> , 2018, 22, 6068-6076.	1.6	5
27	Detection of a heterozygous germline APC mutation in a three-generation family with familial adenomatous polyposis using targeted massive parallel sequencing in Vietnam. <i>BMC Medical Genetics</i> , 2018, 19, 188.	2.1	4
28	ClinPred: Prediction Tool to Identify Disease-Relevant Nonsynonymous Single-Nucleotide Variants. <i>American Journal of Human Genetics</i> , 2018, 103, 474-483.	2.6	149
29	The Ancestral Pace of Variant Reclassification. <i>Journal of the National Cancer Institute</i> , 2018, 110, 1133-1134.	3.0	7
30	GWAS4D: multidimensional analysis of context-specific regulatory variant for human complex diseases and traits. <i>Nucleic Acids Research</i> , 2018, 46, W114-W120.	6.5	69
31	LitVar: a semantic search engine for linking genomic variant data in PubMed and PMC. <i>Nucleic Acids Research</i> , 2018, 46, W530-W536.	6.5	96
32	Dynamics and Thermodynamics of Transthyretin Association from Molecular Dynamics Simulations. <i>BioMed Research International</i> , 2018, 2018, 1-14.	0.9	9
33	In-frame de novo mutation in <i>BICD2</i> in two patients with muscular atrophy and arthrogryposis. <i>Journal of Physical Education and Sports Management</i> , 2018, 4, a003160.	0.5	14
34	Identification of mutations in the <i>PARK2</i> gene in Serbian patients with Parkinson's disease. <i>Journal of the Neurological Sciences</i> , 2018, 393, 27-30.	0.3	8
35	<i>CDKN2A</i> Germline Rare Coding Variants and Risk of Pancreatic Cancer in Minority Populations. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2018, 27, 1364-1370.	1.1	23
36	Expressed HNSCC variants by HPV-status in a well-characterized Michigan cohort. <i>Scientific Reports</i> , 2018, 8, 11458.	1.6	18

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37	Two Angelman families with unusually advanced neurodevelopment carry a start codon variant in the most highly expressed <i>UBE3A</i> isoform. American Journal of Medical Genetics, Part A, 2018, 176, 1641-1647.	0.7	17
38	hgvs: A Python package for manipulating sequence variants using HGVS nomenclature: 2018 Update. Human Mutation, 2018, 39, 1803-1813.	1.1	20
39	Comprehensive screening shows that mutations in the known syndromic genes are rare in infants presenting with hyperinsulinaemic hypoglycaemia. Clinical Endocrinology, 2018, 89, 621-627.	1.2	5
40	Research update for articles published in <i>EJCI</i> in 2016. European Journal of Clinical Investigation, 2018, 48, e13016.	1.7	0
41	Determining the Pathogenicity of a Genomic Variant of Uncertain Significance Using CRISPR/Cas9 and Human-Induced Pluripotent Stem Cells. Circulation, 2018, 138, 2666-2681.	1.6	112
42	Mouse models as a tool for discovering new neurological diseases. Neurobiology of Learning and Memory, 2019, 165, 106902.	1.0	17
43	Report of a bi-allelic truncating germline mutation in TP53. Familial Cancer, 2019, 18, 101-104.	0.9	3
44	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
45	Prenatal chromosomal microarray testing of fetuses with ultrasound structural anomalies: A prospective cohort study of over 1000 consecutive cases. Prenatal Diagnosis, 2019, 39, 1064-1069.	1.1	16
46	Haploinsufficiency in the ANKS1B gene encoding AIDA-1 leads to a neurodevelopmental syndrome. Nature Communications, 2019, 10, 3529.	5.8	20
47	Proteogenomic landscape of squamous cell lung cancer. Nature Communications, 2019, 10, 3578.	5.8	84
48	Atomic Mechanisms of Timothy Syndrome-Associated Mutations in Calcium Channel Cav1.2. Frontiers in Physiology, 2019, 10, 335.	1.3	11
49	Multimodality Imaging of Danon Disease in a Patient with a Novel LAMP2 Mutation. Case, 2019, 3, 235-238.	0.1	0
50	<i>MAGEL2</i> -related disorders: A study and case series. Clinical Genetics, 2019, 96, 493-505.	1.0	26
52	Unusual β^2 -Globin Haplotype Distribution in Newborns from Bengo, Angola. Hemoglobin, 2019, 43, 149-154.	0.4	5
53	An actionable KCNH2 Long QT Syndrome variant detected by sequence and haplotype analysis in a population research cohort. Scientific Reports, 2019, 9, 10964.	1.6	17
54	Deciphering exome sequencing data: Bringing mitochondrial DNA variants to light. Human Mutation, 2019, 40, 2430-2443.	1.1	11
55	Functional analysis of clinical BARD1 germline variants. Journal of Physical Education and Sports Management, 2019, 5, a004093.	0.5	6

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56	Structural Mapping of Missense Mutations in the Pex1/Pex6 Complex. <i>International Journal of Molecular Sciences</i> , 2019, 20, 3756.	1.8	15
57	Molecular diagnostics of Mendelian disorders via combined DNA and RNA sequencing. <i>Medizinische Genetik</i> , 2019, 31, 191-197.	0.1	0
58	Whole exome and targeted gene sequencing to detect pathogenic recessive variants in early onset cerebellar ataxia. <i>Clinical Genetics</i> , 2019, 96, 566-574.	1.0	18
59	Molecular characterization of gastric-type endocervical adenocarcinoma using next-generation sequencing. <i>Modern Pathology</i> , 2019, 32, 1823-1833.	2.9	52
60	Age-adjusted association of homologous recombination genes with ovarian cancer using clinical exomes as controls. <i>Hereditary Cancer in Clinical Practice</i> , 2019, 17, 19.	0.6	7
61	Assessing predictions on fitness effects of missense variants in calmodulin. <i>Human Mutation</i> , 2019, 40, 1463-1473.	1.1	8
62	CAGI SickKids challenges: Assessment of phenotype and variant predictions derived from clinical and genomic data of children with undiagnosed diseases. <i>Human Mutation</i> , 2019, 40, 1373-1391.	1.1	10
63	Contribution of New Adenomatous Polyposis Predisposition Genes in an Unexplained Attenuated Spanish Cohort by Multigene Panel Testing. <i>Scientific Reports</i> , 2019, 9, 9814.	1.6	9
64	Allelic variants of breast cancer susceptibility genes PALB2 and RECQL in the Latvian population. <i>Hereditary Cancer in Clinical Practice</i> , 2019, 17, 17.	0.6	2
65	Clinical Utility of GliSeq Next-Generation Sequencing Test in Pediatric and Young Adult Patients With Brain Tumors. <i>Journal of Neuropathology and Experimental Neurology</i> , 2019, 78, 694-702.	0.9	3
66	Computational assessment of somatic and germline mutations of p16INK4a: Structural insights and implications in disease. <i>Informatics in Medicine Unlocked</i> , 2019, 17, 100208.	1.9	2
67	A novel ISCA2 variant responsible for an early-onset neurodegenerative mitochondrial disorder: a case report of multiple mitochondrial dysfunctions syndrome 4. <i>BMC Neurology</i> , 2019, 19, 153.	0.8	10
68	<i>KCNC1</i> -related disorders: new de novo variants expand the phenotypic spectrum. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1319-1326.	1.7	43
69	Systematic Review of Somatic Mutations in Splenic Marginal Zone Lymphoma. <i>Scientific Reports</i> , 2019, 9, 10444.	1.6	23
70	Haploinsufficiency of the Notch Ligand DLL1 Causes Variable Neurodevelopmental Disorders. <i>American Journal of Human Genetics</i> , 2019, 105, 631-639.	2.6	42
71	Quantifying gene selection in cancer through protein functional alteration bias. <i>Nucleic Acids Research</i> , 2019, 47, 6642-6655.	6.5	21
72	Alagille syndrome mutation update: Comprehensive overview of <i>JAG1</i> and <i>NOTCH2</i> mutation frequencies and insight into missense variant classification. <i>Human Mutation</i> , 2019, 40, 2197-2220.	1.1	84
73	Functional analysis of novel <i>RUNX2</i> mutations identified in patients with cleidocranial dysplasia. <i>Clinical Genetics</i> , 2019, 96, 429-438.	1.0	17

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75	Expression of Normally Repressed Myosin Heavy Chain 7b in the Mammalian Heart Induces Dilated Cardiomyopathy. <i>Journal of the American Heart Association</i> , 2019, 8, e013318.	1.6	16
76	Characterization of a novel SCN5A genetic variant A1294G associated with mixed clinical phenotype. <i>Biochemical and Biophysical Research Communications</i> , 2019, 516, 777-783.	1.0	8
77	Clinical genetic testing in endocrinology: Current concepts and contemporary challenges. <i>Clinical Endocrinology</i> , 2019, 91, 587-607.	1.2	17
78	Eli Nathans. Peter von Zahn's Cold War Broadcasts to West Germany: Assessing America. <i>American Historical Review</i> , 2019, 124, 1539-1540.	0.0	0
79	Bayesian modeling to predict malignant hyperthermia susceptibility and pathogenicity of <i>RYR1</i> , <i>CACNA1S</i> and <i>STAC3</i> variants. <i>Pharmacogenomics</i> , 2019, 20, 989-1003.	0.6	1
80	Spectrum of K _V 2.1 Dysfunction in <i>KCNB1</i> -Associated Neurodevelopmental Disorders. <i>Annals of Neurology</i> , 2019, 86, 899-912.	2.8	52
81	HaTSPIL: A modular pipeline for high-throughput sequencing data analysis. <i>PLoS ONE</i> , 2019, 14, e0222512.	1.1	4
82	A virtual molecular tumor board to improve efficiency and scalability of delivering precision oncology to physicians and their patients. <i>JAMIA Open</i> , 2019, 2, 505-515.	1.0	56
83	Mechanism of Action of Non-Synonymous Single Nucleotide Variations Associated with \pm -Carbonic Anhydrase II Deficiency. <i>Molecules</i> , 2019, 24, 3987.	1.7	18
84	The DisGeNET knowledge platform for disease genomics: 2019 update. <i>Nucleic Acids Research</i> , 2020, 48, D845-D855.	6.5	1,083
85	CancerTracer: a curated database for inpatient tumor heterogeneity. <i>Nucleic Acids Research</i> , 2019, 48, D797-D806.	6.5	9
86	Jointly integrating VCF-based variants and OWL-based biomedical ontologies in MongoDB. <i>IEEE/ACM Transactions on Computational Biology and Bioinformatics</i> , 2019, 17, 1-1.	1.9	7
88	Rare Genetic Variants Associated With Sudden Cardiac Death in Adults. <i>Journal of the American College of Cardiology</i> , 2019, 74, 2623-2634.	1.2	27
89	Clinical and ultrastructural findings in an ataxic variant of Kufor-Rakeb syndrome. <i>Folia Neuropathologica</i> , 2019, 57, 285-294.	0.5	9
90	CAUSALdb: a database for disease/trait causal variants identified using summary statistics of genome-wide association studies. <i>Nucleic Acids Research</i> , 2019, 48, D807-D816.	6.5	34
91	Ensembl 2020. <i>Nucleic Acids Research</i> , 2020, 48, D682-D688.	6.5	1,076
93	SAAVpedia: Identification, Functional Annotation, and Retrieval of Single Amino Acid Variants for Proteogenomic Interpretation. <i>Journal of Proteome Research</i> , 2019, 18, 4133-4142.	1.8	1

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94	Genome-wide microhomologies enable precise template-free editing of biologically relevant deletion mutations. <i>Nature Communications</i> , 2019, 10, 4856.	5.8	22
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96	The use of PanDrugs to prioritize anticancer drug treatments in a case of T-ALL based on individual genomic data. <i>BMC Cancer</i> , 2019, 19, 1005.	1.1	5
97	Different mutation profiles between epithelium and stroma in endometriosis and normal endometrium. <i>Human Reproduction</i> , 2019, 34, 1899-1905.	0.4	37
98	Two Novel NF1 Pathogenic Variants Causing the Creation of a New Splice Site in Patients With Neurofibromatosis Type I. <i>Frontiers in Genetics</i> , 2019, 10, 762.	1.1	5
99	A novel homozygous KCNQ3 loss-of-function variant causes non-syndromic intellectual disability and neonatal-onset pharmacodependent epilepsy. <i>Epilepsia Open</i> , 2019, 4, 464-475.	1.3	29
100	Integrative transcriptome imputation reveals tissue-specific and shared biological mechanisms mediating susceptibility to complex traits. <i>Nature Communications</i> , 2019, 10, 3834.	5.8	68
101	Pediatric Cancer Variant Pathogenicity Information Exchange (PeCanPIE): a cloud-based platform for curating and classifying germline variants. <i>Genome Research</i> , 2019, 29, 1555-1565.	2.4	28
102	Biomarker immunoprofile and molecular characteristics in salivary duct carcinoma: clinicopathological and prognostic implications. <i>Human Pathology</i> , 2019, 93, 37-47.	1.1	27
103	The Impact of Genetic Variants on PTEN Molecular Functions and Cellular Phenotypes. <i>Cold Spring Harbor Perspectives in Medicine</i> , 2019, 9, a036228.	2.9	9
104	Exome sequencing of Saudi Arabian patients with ADPKD. <i>Renal Failure</i> , 2019, 41, 842-849.	0.8	6
105	Low DHEAS Concentration in a Girl Presenting with Short Stature and Premature Pubarche: A Novel <i>PAPSS2</i> Gene Mutation. <i>Hormone Research in Paediatrics</i> , 2019, 92, 262-268.	0.8	6
106	Determining the pathogenicity of CFTR missense variants: Multiple comparisons of in silico predictors and variant annotation databases. <i>Genetics and Molecular Biology</i> , 2019, 42, 560-570.	0.6	6
107	regBase: whole genome base-wise aggregation and functional prediction for human non-coding regulatory variants. <i>Nucleic Acids Research</i> , 2019, 47, e134-e134.	6.5	41
108	Pathogenic APC Variants in Latvian Familial Adenomatous Polyposis Patients. <i>Medicina (Lithuania)</i> , 2019, 55, 612.	0.8	1
109	Next-generation sequencing in Charcot-Marie-Tooth disease: opportunities and challenges. <i>Nature Reviews Neurology</i> , 2019, 15, 644-656.	4.9	140
110	Myosin motor domains carrying mutations implicated in early or late onset hypertrophic cardiomyopathy have similar properties. <i>Journal of Biological Chemistry</i> , 2019, 294, 17451-17462.	1.6	26
111	Functional interpretation of genetic variants using deep learning predicts impact on chromatin accessibility and histone modification. <i>Nucleic Acids Research</i> , 2019, 47, 10597-10611.	6.5	39

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112	Mexican BRCA1 founder mutation: Shortening the gap in genetic assessment for hereditary breast and ovarian cancer patients. <i>PLoS ONE</i> , 2019, 14, e0222709.	1.1	10
113	A 14-year-old in heart failure with multiple cardiomyopathy variants illustrates a role for signal-to-noise analysis in gene test re-interpretation. <i>Clinical Case Reports (discontinued)</i> , 2019, 7, 211-217.	0.2	9
114	Biophysical and Mechanistic Models for Disease-Causing Protein Variants. <i>Trends in Biochemical Sciences</i> , 2019, 44, 575-588.	3.7	143
115	The application of precision medicine in diagnosing familial Mediterranean fever. <i>Leukemia and Lymphoma</i> , 2019, 60, 2091-2093.	0.6	0
116	HuVarBase: A human variant database with comprehensive information at gene and protein levels. <i>PLoS ONE</i> , 2019, 14, e0210475.	1.1	29
117	The ReproGenomics Viewer: a multi-omics and cross-species resource compatible with single-cell studies for the reproductive science community. <i>Bioinformatics</i> , 2019, 35, 3133-3139.	1.8	49
118	Next-generation sequencing of 32 genes associated with hereditary aortopathies and related disorders of connective tissue in a cohort of 199 patients. <i>Genetics in Medicine</i> , 2019, 21, 1832-1841.	1.1	26
119	Whole Exome Sequencing Identifies a Novel Pathogenic RET Variant in Hirschsprung Disease. <i>Frontiers in Genetics</i> , 2018, 9, 752.	1.1	12
120	Associations between single nucleotide polymorphisms and erythrocyte parameters in humans: A systematic literature review. <i>Mutation Research - Reviews in Mutation Research</i> , 2019, 779, 58-67.	2.4	10
121	An interaction-based model for neuropsychiatric features of copy-number variants. <i>PLoS Genetics</i> , 2019, 15, e1007879.	1.5	39
122	A Vietnamese human genetic variation database. <i>Human Mutation</i> , 2019, 40, 1664-1675.	1.1	36
123	Investigation of new candidate genes in retinoblastoma using the TruSight One "clinical exome" gene panel. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e785.	0.6	7
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125	Optimizing Genetic Workup in Pheochromocytoma and Paraganglioma by Integrating Diagnostic and Research Approaches. <i>Cancers</i> , 2019, 11, 809.	1.7	23
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127	Analysis of enriched rare variants in JPH2-encoded junctophilin-2 among Greater Middle Eastern individuals reveals a novel homozygous variant associated with neonatal dilated cardiomyopathy. <i>Scientific Reports</i> , 2019, 9, 9038.	1.6	22
128	Assessing the performance of in silico methods for predicting the pathogenicity of variants in the gene CHEK2, among Hispanic females with breast cancer. <i>Human Mutation</i> , 2019, 40, 1612-1622.	1.1	8
129	Clear cell renal cell carcinoma with Paneth-like cells: Clinicopathologic, morphologic, immunohistochemical, ultrastructural, and molecular analysis of 13 cases. <i>Annals of Diagnostic Pathology</i> , 2019, 41, 96-101.	0.6	5

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130	A case study of a long-term glioblastoma survivor with unmethylated MGMT and hypermutated genotype. <i>Journal of Physical Education and Sports Management</i> , 2019, 5, a003251.	0.5	2
131	First Responder to Genomic Information: A Guide for Primary Care Providers. <i>Molecular Diagnosis and Therapy</i> , 2019, 23, 459-466.	1.6	7
132	When you're strange: Unusual features of the MUTYH glycosylase and implications in cancer. <i>DNA Repair</i> , 2019, 80, 16-25.	1.3	27
133	A Review of the Genomic Analysis of Children Presenting with Developmental Delay/Intellectual Disability and Associated Dysmorphic Features. <i>Cureus</i> , 2019, 11, e3873.	0.2	11
134	A Deep Neural Network for Predicting and Engineering Alternative Polyadenylation. <i>Cell</i> , 2019, 178, 91-106.e23.	13.5	141
135	MTR-Viewer: identifying regions within genes under purifying selection. <i>Nucleic Acids Research</i> , 2019, 47, W121-W126.	6.5	43
136	The road map of cancer precision medicine with the innovation of advanced cancer detection technology and personalized immunotherapy. <i>Japanese Journal of Clinical Oncology</i> , 2019, 49, 596-603.	0.6	10
137	Carrier screening for recessive disorders. <i>Nature Reviews Genetics</i> , 2019, 20, 549-561.	7.7	84
138	BARD1 is a Low/Moderate Breast Cancer Risk Gene: Evidence Based on an Association Study of the Central European p.Q564X Recurrent Mutation. <i>Cancers</i> , 2019, 11, 740.	1.7	25
139	Expression and function of nesfatin-1 are altered by stage of the estrous cycle. <i>American Journal of Physiology - Regulatory Integrative and Comparative Physiology</i> , 2019, 317, R328-R336.	0.9	3
140	WDSPdb: an updated resource for WD40 proteins. <i>Bioinformatics</i> , 2019, 35, 4824-4826.	1.8	18
141	Phenotype characterisation of <i>TBX4</i> mutation and deletion carriers with neonatal and paediatric pulmonary hypertension. <i>European Respiratory Journal</i> , 2019, 54, 1801965.	3.1	77
142	Analysis of hereditary cancer syndromes by using a panel of genes: novel and multiple pathogenic mutations. <i>BMC Cancer</i> , 2019, 19, 535.	1.1	77
143	Efficient and precise base editing in rabbits using human APOBEC3A-nCas9 fusions. <i>Cell Discovery</i> , 2019, 5, 31.	3.1	22
144	A pan-cancer analysis of synonymous mutations. <i>Nature Communications</i> , 2019, 10, 2569.	5.8	147
145	<i>CYP24A1</i> and <i>SLC34A1</i> genetic defects associated with idiopathic infantile hypercalcemia: from genotype to phenotype. <i>Clinical Chemistry and Laboratory Medicine</i> , 2019, 57, 1650-1667.	1.4	30
146	VarMap: a web tool for mapping genomic coordinates to protein sequence and structure and retrieving protein structural annotations. <i>Bioinformatics</i> , 2019, 35, 4854-4856.	1.8	46
147	Assessment of Potential Clinical Role for Exome Sequencing in Schizophrenia. <i>Schizophrenia Bulletin</i> , 2020, 46, 328-335.	2.3	7

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148	Good Intentions Gone Bad. <i>Circulation Genomic and Precision Medicine</i> , 2019, 12, e002560.	1.6	0
149	Simple ClinVar: an interactive web server to explore and retrieve gene and disease variants aggregated in ClinVar database. <i>Nucleic Acids Research</i> , 2019, 47, W99-W105.	6.5	51
150	Gene pathogenicity prediction of Mendelian diseases via the random forest algorithm. <i>Human Genetics</i> , 2019, 138, 673-679.	1.8	4
151	GCK-MODY in the US Monogenic Diabetes Registry: Description of 27 unpublished variants. <i>Diabetes Research and Clinical Practice</i> , 2019, 151, 231-236.	1.1	14
152	Improved base editor for efficient editing in GC contexts in rabbits with an optimized AID-Cas9 fusion. <i>FASEB Journal</i> , 2019, 33, 9210-9219.	0.2	26
153	Navigating the non-coding genome in heart development and Congenital Heart Disease. <i>Differentiation</i> , 2019, 107, 11-23.	1.0	17
154	Diagnostic high-throughput sequencing of 2396 patients with bleeding, thrombotic, and platelet disorders. <i>Blood</i> , 2019, 134, 2082-2091.	0.6	131
155	Beta-Catenin Mutation with Complex Chromosomal Changes in Desmoid Tumor of the Scalp: A Case Report. <i>Craniofacial Trauma & Reconstruction</i> , 2019, 12, 146-149.	0.6	1
156	Examination of rare genetic variants in dental enamel genes: The potential role of next-generation sequencing in primary dental care. <i>Orthodontics and Craniofacial Research</i> , 2019, 22, 49-55.	1.2	1
157	Genetic Variants Are Not Rare in ICD Candidates with Dilated Cardiomyopathy: Time for Next-Generation Sequencing?. <i>Cardiology Research and Practice</i> , 2019, 2019, 1-9.	0.5	3
158	Can Predicted Protein 3D Structures Provide Reliable Insights into whether Missense Variants Are Disease Associated?. <i>Journal of Molecular Biology</i> , 2019, 431, 2197-2212.	2.0	344
159	BRCA1/2 Variant Data-Sharing Practices. <i>Journal of Law, Medicine and Ethics</i> , 2019, 47, 88-96.	0.4	4
160	An overview of the genetic basis of epidermolysis bullosa in Brazil: discovery of novel and recurrent disease-causing variants. <i>Clinical Genetics</i> , 2019, 96, 189-198.	1.0	22
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1106	Functional annotation of de novo variants from healthy individuals. <i>Genomics and Informatics</i> , 2019, 17, e46.	0.4	1
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