David N Cooper

List of Publications by Year in descending order

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469	83,715	2423	528
papers	citations	97 h-index	266 g-index

523	523	523	95086
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A global reference for human genetic variation. Nature, 2015, 526, 68-74.	13.7	13,998
2	Analysis of protein-coding genetic variation in 60,706 humans. Nature, 2016, 536, 285-291.	13.7	9,051
3	A map of human genome variation from population-scale sequencing. Nature, 2010, 467, 1061-1073.	13.7	7,209
4	An integrated map of genetic variation from 1,092 human genomes. Nature, 2012, 491, 56-65.	13.7	7,199
5	MutationTaster2: mutation prediction for the deep-sequencing age. Nature Methods, 2014, 11, 361-362.	9.0	3,203
6	Genome sequence of the Brown Norway rat yields insights into mammalian evolution. Nature, 2004, 428, 493-521.	13.7	1,943
7	Human Gene Mutation Database (HGMDÂ®): 2003 update. Human Mutation, 2003, 21, 577-581.	1.1	1,571
8	Evolutionary and Biomedical Insights from the Rhesus Macaque Genome. Science, 2007, 316, 222-234.	6.0	1,283
9	The mutational spectrum of single base-pair substitutions in mRNA splice junctions of human genes: Causes and consequences. Human Genetics, 1992, 90, 41-54.	1.8	1,182
10	The Human Gene Mutation Database: building a comprehensive mutation repository for clinical and molecular genetics, diagnostic testing and personalized genomic medicine. Human Genetics, 2014, 133, 1-9.	1.8	1,153
11	The Human Gene Mutation Database: towards a comprehensive repository of inherited mutation data for medical research, genetic diagnosis and next-generation sequencing studies. Human Genetics, 2017, 136, 665-677.	1.8	1,106
12	A Systematic Survey of Loss-of-Function Variants in Human Protein-Coding Genes. Science, 2012, 335, 823-828.	6.0	1,095
13	Predicting the Functional, Molecular, and Phenotypic Consequences of Amino Acid Substitutions using Hidden Markov Models. Human Mutation, 2013, 34, 57-65.	1.1	1,057
14	Mapping copy number variation by population-scale genome sequencing. Nature, 2011, 470, 59-65.	13.7	991
15	The CpG dinucleotide and human genetic disease. Human Genetics, 1988, 78, 151-155.	1.8	932
16	The Human Gene Mutation Database: 2008 update. Genome Medicine, 2009, 1, 13.	3.6	774
17	The yak genome and adaptation to life at high altitude. Nature Genetics, 2012, 44, 946-949.	9.4	708
18	Automated inference of molecular mechanisms of disease from amino acid substitutions. Bioinformatics, 2009, 25, 2744-2750.	1.8	691

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19	Insights into hominid evolution from the gorilla genome sequence. Nature, 2012, 483, 169-175.	13.7	663
20	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
21	Demographic history and rare allele sharing among human populations. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 11983-11988.	3.3	589
22	Gene conversion: mechanisms, evolution and human disease. Nature Reviews Genetics, 2007, 8, 762-775.	7.7	576
23	Where genotype is not predictive of phenotype: towards an understanding of the molecular basis of reduced penetrance in human inherited disease. Human Genetics, 2013, 132, 1077-1130.	1.8	528
24	Variation in genome-wide mutation rates within and between human families. Nature Genetics, 2011, 43, 712-714.	9.4	525
25	An integrative approach to predicting the functional effects of non-coding and coding sequence variation. Bioinformatics, 2015, 31, 1536-1543.	1.8	524
26	Gene expression across mammalian organ development. Nature, 2019, 571, 505-509.	13.7	490
27	Gene deletions causing human genetic disease: mechanisms of mutagenesis and the role of the local DNA sequence environment. Human Genetics, 1991, 86, 425-41.	1.8	438
28	Identifying Mendelian disease genes with the Variant Effect Scoring Tool. BMC Genomics, 2013, 14, S3.	1.2	360
29	The mutational spectrum of single base-pair substitutions causing human genetic disease: patterns and predictions. Human Genetics, 1990, 85, 55-74.	1.8	358
30	The Human Gene Mutation Database (HGMDÂ®): optimizing its use in a clinical diagnostic or research setting. Human Genetics, 2020, 139, 1197-1207.	1.8	353
31	A meta-analysis of nonsense mutations causing human genetic disease. Human Mutation, 2008, 29, 1037-1047.	1.1	348
32	Mountain gorilla genomes reveal the impact of long-term population decline and inbreeding. Science, 2015, 348, 242-245.	6.0	326
33	Single base-pair substitutions in exon-intron junctions of human genes: nature, distribution, and consequences for mRNA splicing. Human Mutation, 2007, 28, 150-158.	1.1	324
34	Inferring the molecular and phenotypic impact of amino acid variants with MutPred2. Nature Communications, 2020, 11, 5918.	5.8	305
35	Cytosine methylation and the fate of CpG dinucleotides in vertebrate genomes. Human Genetics, 1989, 83, 181-188.	1.8	303
36	An estimate of unique DNA sequence heterozygosity in the human genome. Human Genetics, 1985, 69, 201-205.	1.8	298

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37	FATHMM-XF: accurate prediction of pathogenic point mutations via extended features. Bioinformatics, 2018, 34, 511-513.	1.8	296
38	Neighboring-Nucleotide Effects on the Rates of Germ-Line Single-Base-Pair Substitution in Human Genes. American Journal of Human Genetics, 1998, 63, 474-488.	2.6	291
39	Splicing factor SFRS1 recognizes a functionally diverse landscape of RNA transcripts. Genome Research, 2009, 19, 381-394.	2.4	284
40	Genome sequencing and comparison of two nonhuman primate animal models, the cynomolgus and Chinese rhesus macaques. Nature Biotechnology, 2011, 29, 1019-1023.	9.4	284
41	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
42	Deleterious- and Disease-Allele Prevalence in Healthy Individuals: Insights from Current Predictions, Mutation Databases, and Population-Scale Resequencing. American Journal of Human Genetics, 2012, 91, 1022-1032.	2.6	255
43	Effect of predicted protein-truncating genetic variants on the human transcriptome. Science, 2015, 348, 666-669.	6.0	252
44	The human gene mutation database. Trends in Genetics, 1997, 13, 121-122.	2.9	249
45	The mutation significance cutoff: gene-level thresholds for variant predictions. Nature Methods, 2016, 13, 109-110.	9.0	249
46	Long-read sequence analysis of the MECP2 gene in Rett syndrome patients: correlation of disease severity with mutation type and location. Human Molecular Genetics, 2000, 9, 1119-1129.	1.4	245
47	Human Gene Mutation Database?A biomedical information and research resource. , 2000, 15, 45-51.		241
48	The human gene mutation database. Nucleic Acids Research, 1998, 26, 285-287.	6.5	231
49	Translocation and gross deletion breakpoints in human inherited disease and cancer I: Nucleotide composition and recombination-associated motifs. Human Mutation, 2003, 22, 229-244.	1.1	214
50	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
51	Predicting the functional consequences of cancer-associated amino acid substitutions. Bioinformatics, 2013, 29, 1504-1510.	1.8	208
52	A systematic analysis of LINE-1 endonuclease-dependent retrotranspositional events causing human genetic disease. Human Genetics, 2005, 117, 411-427.	1.8	206
53	The NF1 somatic mutational landscape in sporadic human cancers. Human Genomics, 2017, 11, 13.	1.4	203
54	Gains of glycosylation comprise an unexpectedly large group of pathogenic mutations. Nature Genetics, 2005, 37, 692-700.	9.4	198

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55	The Human Gene Mutation Database (HGMD) and Its Exploitation in the Fields of Personalized Genomics and Molecular Evolution. Current Protocols in Bioinformatics, 2012, 39, Unit1.13.	25.8	198
56	Eukaryotic DNA methylation. Human Genetics, 1983, 64, 315-333.	1.8	193
57	Functional intronic polymorphisms: Buried treasure awaiting discovery within our genes. Human Genomics, 2010, 4, 284.	1.4	192
58	Breakpoints of gross deletions coincide with non-B DNA conformations. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 14162-14167.	3.3	184
59	Unmethlated domains in vertebrate DNA. Nucleic Acids Research, 1983, 11, 647-658.	6.5	174
60	The functional spectrum of low-frequency coding variation. Genome Biology, 2011, 12, R84.	13.9	173
61	Gross deletions of the neurofibromatosis type 1 (NF1) gene are predominantly of maternal origin and commonly associated with a learning disability, dysmorphic features and developmental delay. Human Genetics, 1998, 102, 591-597.	1.8	171
62	Ranking non-synonymous single nucleotide polymorphisms based on disease concepts. Human Genomics, 2014, 8, 11.	1.4	163
63	Emerging genotypeâ€“phenotype relationships in patients with large NF1 deletions. Human Genetics, 2017, 136, 349-376.	1.8	163
64	Genes, mutations, and human inherited disease at the dawn of the age of personalized genomics. Human Mutation, 2010, 31, 631-655.	1.1	161
65	Early onset seizures and Rett-like features associated with mutations in CDKL5. European Journal of Human Genetics, 2005, 13, 1113-1120.	1.4	160
66	Exome sequencing: Dual role as a discovery and diagnostic tool. Annals of Neurology, 2012, 71, 5-14.	2.8	157
67	Loss of exon identity is a common mechanism of human inherited disease. Genome Research, 2011, 21, 1563-1571.	2.4	156
68	DNA restriction fragment length polymorphisms and heterozygosity in the human genome. Human Genetics, 1984, 66, 1-16.	1.8	155
69	Uganda Genome Resource Enables Insights into Population History and Genomic Discovery in Africa. Cell, 2019, 179, 984-1002.e36.	13.5	152
70	The Human Gene Mutation Database: providing a comprehensive central mutation database for molecular diagnostics and personalised genomics. Human Genomics, 2009, 4, 69.	1.4	151
71	The molecular genetics of growth hormone deficiency. Human Genetics, 1998, 103, 255-272.	1.8	148
72	Metaâ€Analysis of gross insertions causing human genetic disease: Novel mutational mechanisms and the role of replication slippage. Human Mutation, 2005, 25, 207-221.	1.1	148

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73	Clinical characterisation of 29 neurofibromatosis type-1 patients with molecularly ascertained 1.4 Mb type-1 NF1 deletions. Journal of Medical Genetics, 2010, 47, 623-630.	1.5	148
74	Genomic rearrangements in inherited disease and cancer. Seminars in Cancer Biology, 2010, 20, 222-233.	4.3	140
75	CRAVAT: cancer-related analysis of variants toolkit. Bioinformatics, 2013, 29, 647-648.	1.8	140
76	Microdeletions and microinsertions causing human genetic disease: common mechanisms of mutagenesis and the role of local DNA sequence complexity. Human Mutation, 2005, 26, 205-213.	1.1	136
77	A systematic analysis of disease-associated variants in the 3â€² regulatory regions of human protein-coding genes I: general principles and overview. Human Genetics, 2006, 120, 1-21.	1.8	135
78	MutPred Splice: machine learning-based prediction of exonic variants that disrupt splicing. Genome Biology, 2014, 15, R19.	13.9	135
79	A systematic analysis of disease-associated variants in the 3â€² regulatory regions of human protein-coding genes II: the importance of mRNA secondary structure in assessing the functionality of 3â€² UTR variants. Human Genetics, 2006, 120, 301-333.	1.8	125
80	Precursor-product relationship between vitellogenin and the yolk proteins as derived from the complete sequence of aXenopusvitellogenin gene. Nucleic Acids Research, 1987, 15, 4737-4760.	6.5	123
81	MutationTaster2021. Nucleic Acids Research, 2021, 49, W446-W451.	6.5	122
82	Mechanisms of insertional mutagenesis in human genes causing genetic disease. Human Genetics, 1991, 87, 409-15.	1.8	119
83	Methylation-mediated deamination of 5-methylcytosine appears to give rise to mutations causing human inherited disease in CpNpG trinucleotides, as well as in CpG dinucleotides. Human Genomics, 2010, 4, 406.	1.4	118
84	Translocation and deletion breakpoints in cancer genomes are associated with potential non-B DNA-forming sequences. Nucleic Acids Research, 2016, 44, 5673-5688.	6.5	117
85	Human Gene Mutations Affecting RNA Processing and Translation. Annals of Medicine, 1993, 25, 11-17.	1.5	116
86	Evolutionary conservation and selection of human disease gene orthologs in the rat and mouse genomes. Genome Biology, 2004, 5, R47.	13.9	116
87	Genomic rearrangements in theCFTRgene: Extensive allelic heterogeneity and diverse mutational mechanisms. Human Mutation, 2004, 23, 343-357.	1.1	115
88	Proposed guidelines for papers describing DNA polymorphism-disease associations. Human Genetics, 2002, 110, 207-208.	1.8	114
89	Human genetic disease caused by de novo mitochondrial-nuclear DNA transfer. Human Genetics, 2003, 112, 303-309.	1.8	114
90	Neurofibromatosis type 1-associated tumours: Their somatic mutational spectrum and pathogenesis. Human Genomics, 2011, 5, 623.	1.4	113

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91	Haemophilia A: database of nucleotide substitutions, deletions, insertions and rearrangements of the factor VIII gene, second edition. Nucleic Acids Research, 1994, 22, 3511-3533.	6.5	112
92	Meta-analysis of indels causing human genetic disease: mechanisms of mutagenesis and the role of local DNA sequence complexity. Human Mutation, 2003, 21, 28-44.	1.1	112
93	Novel mutations of the growth hormone 1 (GH1) gene disclosed by modulation of the clinical selection criteria for individuals with short stature. Human Mutation, 2003, 21, 424-440.	1.1	106
94	Mosaic type-1 NF1 microdeletions as a cause of both generalized and segmental neurofibromatosis type-1 (NF1). Human Mutation, 2011, 32, 213-219.	1.1	106
95	The molecular pathogenesis of schwannomatosis, a paradigm for the co-involvement of multiple tumour suppressor genes in tumorigenesis. Human Genetics, 2017, 136, 129-148.	1.8	106
96	Mutational and functional analysis of the neurofibromatosis type 1 (NF1) gene. Human Genetics, 1996, 99, 88-92.	1.8	105
97	Inherited Factor VII Deficiency: Molecular Genetics and Pathophysiology. Thrombosis and Haemostasis, 1997, 78, 151-160.	1.8	103
98	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
99	Human growth hormone 1 (GH1) gene expression: Complex haplotype-dependent influence of polymorphic variation in the proximal promoter and locus control region. Human Mutation, 2003, 21, 408-423.	1.1	99
100	On the sequence-directed nature of human gene mutation: The role of genomic architecture and the local DNA sequence environment in mediating gene mutations underlying human inherited disease. Human Mutation, 2011, 32, 1075-1099.	1.1	99
101	DNA polymorphism and the study of disease associations. Human Genetics, 1988, 78, 299-312.	1.8	98
102	Translocation and gross deletion breakpoints in human inherited disease and cancer II: Potential involvement of repetitive sequence elements in secondary structure formation between DNA ends. Human Mutation, 2003, 22, 245-251.	1.1	98
103	Understanding the recent evolution of the human genome: insights from human-chimpanzee genome comparisons. Human Mutation, 2007, 28, 99-130.	1.1	98
104	Characterization of the somatic mutational spectrum of the neurofibromatosis type 1 (NF1) gene in neurofibromatosis patients with benign and malignant tumors. Human Mutation, 2004, 23, 134-146.	1.1	97
105	mutation3D: Cancer Gene Prediction Through Atomic Clustering of Coding Variants in the Structural Proteome. Human Mutation, 2016, 37, 447-456.	1.1	94
106	Identification and characterization of 15 novel GALC gene mutations causing Krabbe disease. Human Mutation, 2010, 31, E1894-E1914.	1.1	93
107	Human Gene Mutation Database: towards a comprehensive central mutation database. Journal of Medical Genetics, 2007, 45, 124-126.	1.5	90
108	A Conservative Assessment of the Major Genetic Causes of Idiopathic Chronic Pancreatitis: Data from a Comprehensive Analysis of PRSS1, SPINK1, CTRC and CFTR Genes in 253 Young French Patients. PLoS ONE, 2013, 8, e73522.	1.1	89

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109	Complex gene rearrangements caused by serial replication slippage. Human Mutation, 2005, 26, 125-134.	1.1	88
110	Human type I hair keratin pseudogene ? hHaA has functional orthologs in the chimpanzee and gorilla: evidence for recent inactivation of the human gene after the Pan-Homo divergence. Human Genetics, 2001, 108, 37-42.	1.8	87
111	Abundance and length of simple repeats in vertebrate genomes are determined by their structural properties. Genome Research, 2008, 18, 1545-1553.	2.4	87
112	Human genetics and genomics a decade after the release of the draft sequence of the human genome. Human Genomics, 2011, 5, 577.	1.4	86
113	A new paradigm emerges from the study of de novo mutations in the context of neurodevelopmental disease. Molecular Psychiatry, 2013, 18, 141-153.	4.1	85
114	Report of the DNA committee and catalogues of cloned and mapped genes, markers formatted for PCR and DNA polymorphisms (Part 1 of 27). Cytogenetic and Genome Research, 1991, 58, 1190-1211.	0.6	78
115	Gross genomic rearrangements involving deletions in the CFTR gene: characterization of six new events from a large cohort of hitherto unidentified cystic fibrosis chromosomes and meta-analysis of the underlying mechanisms. European Journal of Human Genetics, 2006, 14, 567-576.	1.4	77
116	SPINK1 , PRSS1 , CTRC , and CFTR Genotypes Influence Disease Onset and Clinical Outcomes in Chronic Pancreatitis. Clinical and Translational Gastroenterology, 2018, 9, e204.	1.3	76
117	Diagnostic Exome Sequencing to Elucidate the Genetic Basis of Likely Recessive Disorders in Consanguineous Families. Human Mutation, 2014, 35, 1203-1210.	1.1	75
118	GWAS: heritability missing in action?. European Journal of Human Genetics, 2010, 18, 859-861.	1.4	74
119	Diagnosis of genetic disease using recombinant DNA. Human Genetics, 1986, 73, 1-11.	1.8	71
120	Molecular analysis of the genotype-phenotype relationship in factor X deficiency. Human Genetics, 2000, 106, 249-257.	1.8	71
121	Evaluation of denaturing high performance liquid chromatography (DHPLC) for the mutational analysis of the neurofibromatosis type 1 (NF1) gene. Human Genetics, 2001, 109, 487-497.	1.8	71
122	Mosaicism in sporadic neurofibromatosis type 1: variations on a theme common to other hereditary cancer syndromes?. Journal of Medical Genetics, 2008, 45, 622-631.	1.5	71
123	Inherited Factor X Deficiency: Molecular Genetics and Pathophysiology. Thrombosis and Haemostasis, 1997, 78, 161-172.	1.8	71
124	Interpreting Secondary Cardiac Disease Variants in an Exome Cohort. Circulation: Cardiovascular Genetics, 2013, 6, 337-346.	5.1	70
125	Using Exome Data to Identify Malignant Hyperthermia Susceptibility Mutations. Anesthesiology, 2013, 119, 1043-1053.	1.3	69
126	Restriction fragment length polymorphisms at the human parathyroid hormone gene locus. Human Genetics, 1984, 67, 428-431.	1.8	68

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127	Gonosomal Mosaicism for a Nonsense Mutation (R1947X) in the NF1 Gene in Segmental Neurofibromatosis Type 1. Journal of Investigative Dermatology, 2005, 125, 463-466.	0.3	68
128	Complex patterns of copy number variation at sites of segmental duplications: an important category of structural variation in the human genome. Human Genetics, 2006, 120, 270-284.	1.8	68
129	Molecular mechanisms of chromosomal rearrangement during primate evolution. Chromosome Research, 2008, 16, 41-56.	1.0	68
130	MuPIT interactive: webserver for mapping variant positions to annotated, interactive 3D structures. Human Genetics, 2013, 132, 1235-1243.	1.8	68
131	Localization of a human heat-shock HSP 70 gene sequence to chromosome 6 and detection of two other loci by somatic-cell hybrid and restriction fragment length polymorphism analysis. Human Genetics, 1987, 75, 123-128.	1.8	66
132	IDUA mutational profiling of a cohort of 102 European patients with mucopolysaccharidosis type I: identification and characterization of 35 novel Î±-L-iduronidase (IDUA) alleles. Human Mutation, 2011, 32, E2189-E2210.	1.1	66
133	Individualized Iterative Phenotyping for Genome-wide Analysis of Loss-of-Function Mutations. American Journal of Human Genetics, 2015, 96, 913-925.	2.6	66
134	Gain-of-glycosylation mutations. Current Opinion in Genetics and Development, 2007, 17, 245-251.	1.5	65
135	Disease-causing mutations in the human genome. European Journal of Pediatrics, 2000, 159, S173-S178.	1.3	64
136	Gene conversion causing human inherited disease: Evidence for involvement of non-B-DNA-forming sequences and recombination-promoting motifs in DNA breakage and repair. Human Mutation, 2009, 30, 1189-1198.	1.1	63
137	DDIG-in: discriminating between disease-associated and neutral non-frameshifting micro-indels. Genome Biology, 2013, 14, R23.	13.9	63
138	SVA retrotransposon insertion-associated deletion represents a novel mutational mechanism underlying large genomic copy number changes with non-recurrent breakpoints. Genome Biology, 2014, 15, R80.	13.9	63
139	Estimating the Efficacy and Efficiency of Cascade Genetic Screening. American Journal of Human Genetics, 2001, 69, 361-370.	2.6	62
140	Intrachromosomal serial replication slippage intransgives rise to diverse genomic rearrangements involving inversions. Human Mutation, 2005, 26, 362-373.	1.1	62
141	Gross rearrangements of the MECP2 gene are found in both classical and atypical Rett syndrome patients. Journal of Medical Genetics, 2005, 43, 451-456.	1.5	62
142	Long homopurine*homopyrimidine sequences are characteristic of genes expressed in brain and the pseudoautosomal region. Nucleic Acids Research, 2006, 34, 2663-2675.	6.5	60
143	Type 2 NF1 Deletions Are Highly Unusual by Virtue of the Absence of Nonallelic Homologous Recombination Hotspots and an Apparent Preference for Female Mitotic Recombination. American Journal of Human Genetics, 2007, 81, 1201-1220.	2.6	60
144	AMELIE speeds Mendelian diagnosis by matching patient phenotype and genotype to primary literature. Science Translational Medicine, 2020, 12, .	5.8	60

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145	DNA methylation and CpG suppression. Cell Differentiation, 1985, 17, 199-205.	1.3	59
146	Microattribution and nanopublication as means to incentivize the placement of human genome variation data into the public domain. Human Mutation, 2012, 33, 1503-1512.	1.1	59
147	No Association Between CELâ€“HYB Hybrid Allele and Chronic Pancreatitis in Asian Populations. Gastroenterology, 2016, 150, 1558-1560.e5.	0.6	59
148	In silico functional profiling of human disease-associated and polymorphic amino acid substitutions. Human Mutation, 2010, 31, 335-346.	1.1	57
149	Haemophilia A: database of nucleotide substitutions, deletions, insertions and rearrangements of the factor VIII gene, second edition. Nucleic Acids Research, 1994, 22, 4851-4868.	6.5	56
150	Somatic spectrum of cancer-associated single basepair substitutions in the TP53 gene is determined mainly by endogenous mechanisms of mutation and by selection. Human Mutation, 1995, 5, 48-57.	1.1	56
151	S-CAP extends pathogenicity prediction to genetic variants that affect RNA splicing. Nature Genetics, 2019, 51, 755-763.	9.4	56
152	Gene synteny comparisons between different vertebrates provide new insights into breakage and fusion events during mammalian karyotype evolution. BMC Evolutionary Biology, 2009, 9, 84.	3.2	54
153	A Novel Dysfunctional Growth Hormone Variant (Ile179Met) Exhibits a Decreased Ability to Activate the Extracellular Signal-Regulated Kinase Pathway. Journal of Clinical Endocrinology and Metabolism, 2004, 89, 1068-1075.	1.8	53
154	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
155	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
156	RegSNPs-intron: a computational framework for predicting pathogenic impact of intronic single nucleotide variants. Genome Biology, 2019, 20, 254.	3.8	52
157	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
158	LINE-1 Endonuclease-Dependent Retrotranspositional Events Causing Human Genetic Disease: Mutation Detection Bias and Multiple Mechanisms of Target Gene Disruption. Journal of Biomedicine and Biotechnology, 2006, 2006, 1-9.	3.0	51
159	Closely spaced multiple mutations as potential signatures of transient hypermutability in human genes. Human Mutation, 2009, 30, 1435-1448.	1.1	51
160	Report of the DNA committee and catalogues of cloned and mapped genes and DNA polymorphisms (Part 1 of 14). Cytogenetic and Genome Research, 1990, 55, 457-472.	0.6	50
161	The Human Gene Mutation Database (HGMD) and Its Exploitation in the Study of Mutational Mechanisms. Current Protocols in Bioinformatics, 2005, 12, 1.13.1-1.13.20.	25.8	50
162	Genotype-phenotype associations in neurofibromatosis type 1 (NF1): an increased risk of tumor complications in patients with NF1splice-site mutations?. Human Genomics, 2012, 6, 12.	1.4	50

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163	Structural divergence between the human and chimpanzee genomes. Human Genetics, 2007, 120, 759-778.	1.8	49
164	Mechanisms of Base Substitution Mutagenesis in Cancer Genomes. Genes, 2014, 5, 108-146.	1.0	49
165	Key challenges for nextâ€generation pharmacogenomics. EMBO Reports, 2014, 15, 472-476.	2.0	49
166	Extensive disruption of protein interactions by genetic variants across the allele frequency spectrum in human populations. Nature Communications, 2019, 10, 4141.	5.8	48
167	The effect of replication errors on the mismatch analysis of PCR-amplified DNA. Nucleic Acids Research, 1990, 18, 973-978.	6.5	47
168	Breakpoint analysis of the pericentric inversion distinguishing human chromosome 4 from the homologous chromosome in the chimpanzee (Pan troglodytes). Human Mutation, 2005, 25, 45-55.	1.1	47
169	A critical view of the general publicâ€™s awareness and physiciansâ€™ opinion of the trends and potential pitfalls of genetic testing in Greece. Personalized Medicine, 2011, 8, 551-561.	0.8	47
170	A Massively Parallel Pipeline to Clone DNA Variants and Examine Molecular Phenotypes of Human Disease Mutations. PLoS Genetics, 2014, 10, e1004819.	1.5	47
171	Critical appraisal of the views of healthcare professionals with respect to pharmacogenomics and personalized medicine in Greece. Personalized Medicine, 2014, 11, 15-26.	0.8	46
172	Developmental Gene Expression Differences between Humans and Mammalian Models. Cell Reports, 2020, 33, 108308.	2.9	46
173	Assessing Radiation-Associated Mutational Risk to the Germline: Repetitive DNA Sequences as Mutational Targets and Biomarkers. Radiation Research, 2006, 165, 249-268.	0.7	45
174	Microarray-based copy number analysis of neurofibromatosis type-1 (NF1)-associated malignant peripheral nerve sheath tumors reveals a role for Rho-GTPase pathway genes in NF1 tumorigenesis. Human Mutation, 2012, 33, 763-776.	1.1	44
175	Genetic tests obtainable through pharmacies: the good, the bad, and the ugly. Human Genomics, 2013, 7, 17.	1.4	44
176	Exome versus transcriptome sequencing in identifying coding region variants. Expert Review of Molecular Diagnostics, 2012, 12, 241-251.	1.5	43
177	Independent intrachromosomal recombination events underlie the pericentric inversions of chimpanzee and gorilla chromosomes homologous to human chromosome 16. Genome Research, 2005, 15, 1232-1242.	2.4	42
178	Mechanisms of Loss of Heterozygosity in Neurofibromatosis Type 1-Associated Plexiform Neurofibromas. Journal of Investigative Dermatology, 2009, 129, 615-621.	0.3	42
179	A novel third type of recurrent NF1 microdeletion mediated by nonallelic homologous recombination between LRRC37B-containing low-copy repeats in 17q11.2. Human Mutation, 2010, 31, 742-751.	1.1	42
180	The genetic structure of the Turkish population reveals high levels of variation and admixture. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	42

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