David N Cooper

List of Publications by Year in descending order

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469	83,715	2322	517
papers	citations	98 h-index	267 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.	27.8	13,998
2	Analysis of protein-coding genetic variation in 60,706 humans. Nature, 2016, 536, 285-291.	27.8	9,051
3	A map of human genome variation from population-scale sequencing. Nature, 2010, 467, 1061-1073.	27.8	7,209
4	An integrated map of genetic variation from 1,092 human genomes. Nature, 2012, 491, 56-65.	27.8	7,199
5	MutationTaster2: mutation prediction for the deep-sequencing age. Nature Methods, 2014, 11, 361-362.	19.0	3,203
6	Genome sequence of the Brown Norway rat yields insights into mammalian evolution. Nature, 2004, 428, 493-521.	27.8	1,943
7	Human Gene Mutation Database (HGMD^{Â®}): 2003 update. Human Mutation, 2003, 21, 577-581.	2.5	1,571
8	Evolutionary and Biomedical Insights from the Rhesus Macaque Genome. Science, 2007, 316, 222-234.	12.6	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.	3.8	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.	3.8	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.	3.8	1,106
12	A Systematic Survey of Loss-of-Function Variants in Human Protein-Coding Genes. Science, 2012, 335, 823-828.	12.6	1,095
13	Predicting the Functional, Molecular, and Phenotypic Consequences of Amino Acid Substitutions using Hidden Markov Models. Human Mutation, 2013, 34, 57-65.	2.5	1,057
14	Mapping copy number variation by population-scale genome sequencing. Nature, 2011, 470, 59-65.	27.8	991
15	The CpG dinucleotide and human genetic disease. Human Genetics, 1988, 78, 151-155.	3.8	932
16	The Human Gene Mutation Database: 2008 update. Genome Medicine, 2009, 1, 13.	8.2	774
17	The yak genome and adaptation to life at high altitude. Nature Genetics, 2012, 44, 946-949.	21.4	708
18	Automated inference of molecular mechanisms of disease from amino acid substitutions. Bioinformatics, 2009, 25, 2744-2750.	4.1	691

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19	Insights into hominid evolution from the gorilla genome sequence. Nature, 2012, 483, 169-175.	27.8	663
20	M-CAP eliminates a majority of variants of uncertain significance in clinical exomes at high sensitivity. Nature Genetics, 2016, 48, 1581-1586.	21.4	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.	7.1	589
22	Gene conversion: mechanisms, evolution and human disease. Nature Reviews Genetics, 2007, 8, 762-775.	16.3	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.	3.8	528
24	Variation in genome-wide mutation rates within and between human families. Nature Genetics, 2011, 43, 712-714.	21.4	525
25	An integrative approach to predicting the functional effects of non-coding and coding sequence variation. Bioinformatics, 2015, 31, 1536-1543.	4.1	524
26	Gene expression across mammalian organ development. Nature, 2019, 571, 505-509.	27.8	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.	3.8	438
28	Identifying Mendelian disease genes with the Variant Effect Scoring Tool. BMC Genomics, 2013, 14, S3.	2.8	360
29	The mutational spectrum of single base-pair substitutions causing human genetic disease: patterns and predictions. Human Genetics, 1990, 85, 55-74.	3.8	358
30	The Human Gene Mutation Database (HGMDÂ®): optimizing its use in a clinical diagnostic or research setting. Human Genetics, 2020, 139, 1197-1207.	3.8	353
31	A meta-analysis of nonsense mutations causing human genetic disease. Human Mutation, 2008, 29, 1037-1047.	2.5	348
32	Mountain gorilla genomes reveal the impact of long-term population decline and inbreeding. Science, 2015, 348, 242-245.	12.6	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.	2.5	324
34	Inferring the molecular and phenotypic impact of amino acid variants with MutPred2. Nature Communications, 2020, 11, 5918.	12.8	305
35	Cytosine methylation and the fate of CpG dinucleotides in vertebrate genomes. Human Genetics, 1989, 83, 181-188.	3.8	303
36	An estimate of unique DNA sequence heterozygosity in the human genome. Human Genetics, 1985, 69, 201-205.	3.8	298

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37	FATHMM-XF: accurate prediction of pathogenic point mutations via extended features. Bioinformatics, 2018, 34, 511-513.	4.1	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.	6.2	291
39	Splicing factor SFRS1 recognizes a functionally diverse landscape of RNA transcripts. Genome Research, 2009, 19, 381-394.	5.5	284
40	Genome sequencing and comparison of two nonhuman primate animal models, the cynomolgus and Chinese rhesus macaques. Nature Biotechnology, 2011, 29, 1019-1023.	17.5	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.	2.5	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.	6.2	255
43	Effect of predicted protein-truncating genetic variants on the human transcriptome. Science, 2015, 348, 666-669.	12.6	252
44	The human gene mutation database. Trends in Genetics, 1997, 13, 121-122.	6.7	249
45	The mutation significance cutoff: gene-level thresholds for variant predictions. Nature Methods, 2016, 13, 109-110.	19.0	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.	2.9	245
47	Human Gene Mutation Database?A biomedical information and research resource. Human Mutation, 2000, 15, 45-51.	2.5	241
48	The human gene mutation database. Nucleic Acids Research, 1998, 26, 285-287.	14.5	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.	2.5	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.	7.1	213
51	Predicting the functional consequences of cancer-associated amino acid substitutions. Bioinformatics, 2013, 29, 1504-1510.	4.1	208
52	A systematic analysis of LINE-1 endonuclease-dependent retrotranspositional events causing human genetic disease. Human Genetics, 2005, 117, 411-427.	3.8	206
53	The NF1 somatic mutational landscape in sporadic human cancers. Human Genomics, 2017, 11, 13.	2.9	203
54	Gains of glycosylation comprise an unexpectedly large group of pathogenic mutations. Nature Genetics, 2005, 37, 692-700.	21.4	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.	3.8	193
57	Functional intronic polymorphisms: Buried treasure awaiting discovery within our genes. Human Genomics, 2010, 4, 284.	2.9	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.	7.1	184
59	Unmethlated domains in vertebrate DNA. Nucleic Acids Research, 1983, 11, 647-658.	14.5	174
60	The functional spectrum of low-frequency coding variation. Genome Biology, 2011, 12, R84.	9.6	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.	3.8	171
62	Ranking non-synonymous single nucleotide polymorphisms based on disease concepts. Human Genomics, 2014, 8, 11.	2.9	163
63	Emerging genotypeâ€“phenotype relationships in patients with large NF1 deletions. Human Genetics, 2017, 136, 349-376.	3.8	163
64	Genes, mutations, and human inherited disease at the dawn of the age of personalized genomics. Human Mutation, 2010, 31, 631-655.	2.5	161
65	Early onset seizures and Rett-like features associated with mutations in CDKL5. European Journal of Human Genetics, 2005, 13, 1113-1120.	2.8	160
66	Exome sequencing: Dual role as a discovery and diagnostic tool. Annals of Neurology, 2012, 71, 5-14.	5.3	157
67	Loss of exon identity is a common mechanism of human inherited disease. Genome Research, 2011, 21, 1563-1571.	5.5	156
68	DNA restriction fragment length polymorphisms and heterozygosity in the human genome. Human Genetics, 1984, 66, 1-16.	3.8	155
69	Uganda Genome Resource Enables Insights into Population History and Genomic Discovery in Africa. Cell, 2019, 179, 984-1002.e36.	28.9	152
70	The Human Gene Mutation Database: providing a comprehensive central mutation database for molecular diagnostics and personalised genomics. Human Genomics, 2009, 4, 69.	2.9	151
71	The molecular genetics of growth hormone deficiency. Human Genetics, 1998, 103, 255-272.	3.8	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.	2.5	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.	3.2	148
74	Genomic rearrangements in inherited disease and cancer. Seminars in Cancer Biology, 2010, 20, 222-233.	9.6	140
75	CRAVAT: cancer-related analysis of variants toolkit. Bioinformatics, 2013, 29, 647-648.	4.1	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.	2.5	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.	3.8	135
78	MutPred Splice: machine learning-based prediction of exonic variants that disrupt splicing. Genome Biology, 2014, 15, R19.	9.6	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.	3.8	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.	14.5	123
81	MutationTaster2021. Nucleic Acids Research, 2021, 49, W446-W451.	14.5	122
82	Mechanisms of insertional mutagenesis in human genes causing genetic disease. Human Genetics, 1991, 87, 409-15.	3.8	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.	2.9	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.	14.5	117
85	Human Gene Mutations Affecting RNA Processing and Translation. Annals of Medicine, 1993, 25, 11-17.	3.8	116
86	Evolutionary conservation and selection of human disease gene orthologs in the rat and mouse genomes. Genome Biology, 2004, 5, R47.	9.6	116
87	Genomic rearrangements in theCFTRgene: Extensive allelic heterogeneity and diverse mutational mechanisms. Human Mutation, 2004, 23, 343-357.	2.5	115
88	Proposed guidelines for papers describing DNA polymorphism-disease associations. Human Genetics, 2002, 110, 207-208.	3.8	114
89	Human genetic disease caused by de novo mitochondrial-nuclear DNA transfer. Human Genetics, 2003, 112, 303-309.	3.8	114
90	Neurofibromatosis type 1-associated tumours: Their somatic mutational spectrum and pathogenesis. Human Genomics, 2011, 5, 623.	2.9	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.	14.5	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.	2.5	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.	2.5	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.	2.5	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.	3.8	106
96	Mutational and functional analysis of the neurofibromatosis type 1 (NF1) gene. Human Genetics, 1996, 99, 88-92.	3.8	105
97	Inherited Factor VII Deficiency: Molecular Genetics and Pathophysiology. Thrombosis and Haemostasis, 1997, 78, 151-160.	3.4	103
98	Assessing the Pathogenicity of Insertion and Deletion Variants with the Variant Effect Scoring Tool (VESTâ€indel). Human Mutation, 2016, 37, 28-35.	2.5	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.	2.5	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.	2.5	99
101	DNA polymorphism and the study of disease associations. Human Genetics, 1988, 78, 299-312.	3.8	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.	2.5	98
103	Understanding the recent evolution of the human genome: insights from human-chimpanzee genome comparisons. Human Mutation, 2007, 28, 99-130.	2.5	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.	2.5	97
105	mutation3D: Cancer Gene Prediction Through Atomic Clustering of Coding Variants in the Structural Proteome. Human Mutation, 2016, 37, 447-456.	2.5	94
106	Identification and characterization of 15 novel GALC gene mutations causing Krabbe disease. Human Mutation, 2010, 31, E1894-E1914.	2.5	93
107	Human Gene Mutation Database: towards a comprehensive central mutation database. Journal of Medical Genetics, 2007, 45, 124-126.	3.2	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.	2.5	89

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109	Complex gene rearrangements caused by serial replication slippage. Human Mutation, 2005, 26, 125-134.	2.5	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.	3.8	87
111	Abundance and length of simple repeats in vertebrate genomes are determined by their structural properties. Genome Research, 2008, 18, 1545-1553.	5.5	87
112	Human genetics and genomics a decade after the release of the draft sequence of the human genome. Human Genomics, 2011, 5, 577.	2.9	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.	7.9	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.	1.1	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.	2.8	77
116	SPINK1 , PRSS1 , CTRC , and CFTR Genotypes Influence Disease Onset and Clinical Outcomes in Chronic Pancreatitis. Clinical and Translational Gastroenterology, 2018, 9, e204.	2.5	76
117	Diagnostic Exome Sequencing to Elucidate the Genetic Basis of Likely Recessive Disorders in Consanguineous Families. Human Mutation, 2014, 35, 1203-1210.	2.5	75
118	GWAS: heritability missing in action?. European Journal of Human Genetics, 2010, 18, 859-861.	2.8	74
119	Diagnosis of genetic disease using recombinant DNA. Human Genetics, 1986, 73, 1-11.	3.8	71
120	Molecular analysis of the genotype-phenotype relationship in factor X deficiency. Human Genetics, 2000, 106, 249-257.	3.8	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.	3.8	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.	3.2	71
123	Inherited Factor X Deficiency: Molecular Genetics and Pathophysiology. Thrombosis and Haemostasis, 1997, 78, 161-172.	3.4	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.	2.5	69
126	Restriction fragment length polymorphisms at the human parathyroid hormone gene locus. Human Genetics, 1984, 67, 428-431.	3.8	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.	0.7	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.	3.8	68
129	Molecular mechanisms of chromosomal rearrangement during primate evolution. Chromosome Research, 2008, 16, 41-56.	2.2	68
130	MuPIT interactive: webserver for mapping variant positions to annotated, interactive 3D structures. Human Genetics, 2013, 132, 1235-1243.	3.8	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.	3.8	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.	2.5	66
133	Individualized Iterative Phenotyping for Genome-wide Analysis of Loss-of-Function Mutations. American Journal of Human Genetics, 2015, 96, 913-925.	6.2	66
134	Gain-of-glycosylation mutations. Current Opinion in Genetics and Development, 2007, 17, 245-251.	3.3	65
135	Disease-causing mutations in the human genome. European Journal of Pediatrics, 2000, 159, S173-S178.	2.7	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.	2.5	63
137	DDIG-in: discriminating between disease-associated and neutral non-frameshifting micro-indels. Genome Biology, 2013, 14, R23.	9.6	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.	9.6	63
139	Estimating the Efficacy and Efficiency of Cascade Genetic Screening. American Journal of Human Genetics, 2001, 69, 361-370.	6.2	62
140	Intrachromosomal serial replication slippage intransgives rise to diverse genomic rearrangements involving inversions. Human Mutation, 2005, 26, 362-373.	2.5	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.	3.2	62
142	Long homopurine*homopyrimidine sequences are characteristic of genes expressed in brain and the pseudoautosomal region. Nucleic Acids Research, 2006, 34, 2663-2675.	14.5	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.	6.2	60
144	AMELIE speeds Mendelian diagnosis by matching patient phenotype and genotype to primary literature. Science Translational Medicine, 2020, 12, .	12.4	60

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145	DNA methylation and CpG suppression. Cell Differentiation, 1985, 17, 199-205.	0.4	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.	2.5	59
147	No Association Between CELâ€“HYB Hybrid Allele and Chronic Pancreatitis in Asian Populations. Gastroenterology, 2016, 150, 1558-1560.e5.	1.3	59
148	In silico functional profiling of human disease-associated and polymorphic amino acid substitutions. Human Mutation, 2010, 31, 335-346.	2.5	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.	14.5	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.	2.5	56
151	S-CAP extends pathogenicity prediction to genetic variants that affect RNA splicing. Nature Genetics, 2019, 51, 755-763.	21.4	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.	3.6	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.	4.1	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.	4.1	52
156	RegSNPs-intron: a computational framework for predicting pathogenic impact of intronic single nucleotide variants. Genome Biology, 2019, 20, 254.	8.8	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.	7.1	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.	2.5	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.	1.1	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.	2.9	50

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163	Structural divergence between the human and chimpanzee genomes. Human Genetics, 2007, 120, 759-778.	3.8	49
164	Mechanisms of Base Substitution Mutagenesis in Cancer Genomes. Genes, 2014, 5, 108-146.	2.4	49
165	Key challenges for nextâ€generation pharmacogenomics. EMBO Reports, 2014, 15, 472-476.	4.5	49
166	Extensive disruption of protein interactions by genetic variants across the allele frequency spectrum in human populations. Nature Communications, 2019, 10, 4141.	12.8	48
167	The effect of replication errors on the mismatch analysis of PCR-amplified DNA. Nucleic Acids Research, 1990, 18, 973-978.	14.5	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.	2.5	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.	1.5	47
170	A Massively Parallel Pipeline to Clone DNA Variants and Examine Molecular Phenotypes of Human Disease Mutations. PLoS Genetics, 2014, 10, e1004819.	3.5	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.	1.5	46
172	Developmental Gene Expression Differences between Humans and Mammalian Models. Cell Reports, 2020, 33, 108308.	6.4	46
173	Assessing Radiation-Associated Mutational Risk to the Germline: Repetitive DNA Sequences as Mutational Targets and Biomarkers. Radiation Research, 2006, 165, 249-268.	1.5	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.	2.5	44
175	Genetic tests obtainable through pharmacies: the good, the bad, and the ugly. Human Genomics, 2013, 7, 17.	2.9	44
176	Exome versus transcriptome sequencing in identifying coding region variants. Expert Review of Molecular Diagnostics, 2012, 12, 241-251.	3.1	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.	5.5	42
178	Mechanisms of Loss of Heterozygosity in Neurofibromatosis Type 1-Associated Plexiform Neurofibromas. Journal of Investigative Dermatology, 2009, 129, 615-621.	0.7	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.	2.5	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, .	7.1	42

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181	Ectopic (Illegitimate) Transcription: New Possibilities for the Analysis and Diagnosis of Human Genetic Disease. Annals of Medicine, 1994, 26, 9-14.	3.8	41
182	Characterisation of a functional intronic polymorphism in the human growth hormone (GHI) gene. Human Genomics, 2010, 4, 289.	2.9	41
183	Deciphering next-generation pharmacogenomics: an information technology perspective. Open Biology, 2014, 4, 140071.	3.6	41
184	Proteins linked to autosomal dominant and autosomal recessive disorders harbor characteristic rare missense mutation distribution patterns. Human Molecular Genetics, 2015, 24, 5995-6002.	2.9	40
185	Assessing the relative importance of the biophysical properties of amino acid substitutions associated with human genetic disease. Human Mutation, 2002, 20, 98-109.	2.5	39
186	Trans-species polymorphism in humans and the great apes is generally maintained by balancing selection that modulates the host immune response. Human Genomics, 2015, 9, 21.	2.9	39
187	Human gene mutation in pathology and evolution. Journal of Inherited Metabolic Disease, 2002, 25, 157-182.	3.6	38
188	Detection of two Alu insertions in the CFTR gene. Journal of Cystic Fibrosis, 2008, 7, 37-43.	0.7	38
189	Molecular characterization of 22 novel UDP-N-acetylglucosamine-1-phosphate transferase Î±- and Î²-subunit (GNPTAB) gene mutations causing mucolipidosis types IIÎ±/Î² and IIIÎ±/Î² in 46 patients. Human Mutation, 2009, 30, E956-E973.	2.5	38
190	Evidence for Cultured Human Vascular Smooth Muscle Cell Heterogeneity: Isolation of Clonal Cells and Study of their Growth Characteristics. Thrombosis and Haemostasis, 1996, 75, 854-858.	3.4	38
191	Molecular genetic analysis of severe proteinÂC deficiency. Human Genetics, 2000, 106, 646-653.	3.8	37
192	Meiotic recombination favors the spreading of deleterious mutations in human populations. Human Mutation, 2011, 32, 198-206.	2.5	37
193	Test Pricing and Reimbursement in Genomic Medicine: Towards a General Strategy. Public Health Genomics, 2016, 19, 352-363.	1.0	37
194	Investigating DNA-, RNA-, and protein-based features as a means to discriminate pathogenic synonymous variants. Human Mutation, 2017, 38, 1336-1347.	2.5	37
195	Chicken lens Î´-crystallin gene expression and methylation in several non-lens tissues. Nucleic Acids Research, 1983, 11, 2513-2527.	14.5	36
196	Intrachromosomal mitotic nonallelic homologous recombination is the major molecular mechanism underlying type-2 NF1 deletions. Human Mutation, 2010, 31, 1163-1173.	2.5	36
197	Identification of large-scale human-specific copy number differences by inter-species array comparative genomic hybridization. Human Genetics, 2006, 119, 185-198.	3.8	35
198	Gene discovery in familial cancer syndromes by exome sequencing: prospects for the elucidation of familial colorectal cancer type X. Modern Pathology, 2012, 25, 1055-1068.	5.5	35

#	Article	IF	Citations
199	New clinical and molecular insights on Barth syndrome. Orphanet Journal of Rare Diseases, 2013, 8, 27.	2.7	35
200	Evolution of the proximal promoter region of the mammalian growth hormone gene. Gene, 1999, 237, 143-151.	2.2	34
201	A meta-analysis of single base-pair substitutions in translational termination codons ('nonstop') Tj ETQq1 1 0.784314 rgBT /Overlock	2.9	34
202	Monozygotic twins discordant for neurofibromatosis type 1 due to a postzygotic NF1 gene mutation. Human Mutation, 2011, 32, E2134-E2147.	2.5	34
203	Patterns and Mutational Signatures of Tandem Base Substitutions Causing Human Inherited Disease. Human Mutation, 2013, 34, 1119-1130.	2.5	34
204	Guanine Holes Are Prominent Targets for Mutation in Cancer and Inherited Disease. PLoS Genetics, 2013, 9, e1003816.	3.5	34
205	An integrative approach to predicting the functional effects of small indels in non-coding regions of the human genome. BMC Bioinformatics, 2017, 18, 442.	2.6	34
206	Pathogenicity and functional impact of non-frameshifting insertion/deletion variation in the human genome. PLoS Computational Biology, 2019, 15, e1007112.	3.2	34
207	De novo splice site mutation in the antithrombin III (AT3) gene causing recurrent venous thrombosis: Demonstration of exon skipping by ectopic transcript analysis. Genomics, 1992, 13, 1359-1361.	2.9	33
208	From the periphery to centre stage: de novo single nucleotide variants play a key role in human genetic disease. Journal of Medical Genetics, 2013, 50, 203-211.	3.2	33
209	Down's syndrome and the molecular biology of chromosome 21. Progress in Neurobiology, 1988, 30, 507-530.	5.7	32
210	Late-onset homozygous protein C deficiency. Lancet, The, 1991, 338, 575-576.	13.7	32
211	Molecular Genetic Analysis of the PLP1 Gene in 38 Families with PLP1-related disorders: Identification and Functional Characterization of 11 Novel PLP1 Mutations. Orphanet Journal of Rare Diseases, 2011, 6, 40.	2.7	32
212	Single base-pair substitutions at the translation initiation sites of human genes as a cause of inherited disease. Human Mutation, 2011, 32, 1137-1143.	2.5	32
213	The emergence of the mitochondrial genome as a partial regulator of nuclear function is providing new insights into the genetic mechanisms underlying age-related complex disease. Human Genetics, 2014, 133, 435-458.	3.8	32
214	Clarifying the clinical relevance ofSPINK1intronic variants in chronic pancreatitis. Gut, 2016, 65, 884-886.	12.1	32
215	Mutational signatures and mutable motifs in cancer genomes. Briefings in Bioinformatics, 2017, 19, 1085-1101.	6.5	32
216	DNA polymerase Î· mutational signatures are found in a variety of different types of cancer. Cell Cycle, 2018, 17, 348-355.	2.6	32

#	Article	IF	Citations
217	Cruciform-forming inverted repeats appear to have mediated many of the microinversions that distinguish the human and chimpanzee genomes. Chromosome Research, 2009, 17, 469-483.	2.2	31
218	Complete ascertainment of intragenic copy number mutations (CNMs) in the CFTR gene and its implications for CNM formation at other autosomal loci. Human Mutation, 2010, 31, 421-428.	2.5	31
219	Non-B DNA-forming Sequences and WRN Deficiency Independently Increase the Frequency of Base Substitution in Human Cells. Journal of Biological Chemistry, 2011, 286, 10017-10026.	3.4	31
220	Detection of missense mutations by single-strand conformational polymorphism (SSCP) analysis in five dysfunctional variants of coagulation factor VII. Human Molecular Genetics, 1993, 2, 1355-1359.	2.9	30
221	Hypermethylation of the neurofibromatosis type 1 (NF1) gene promoter is not a common event in the inactivation of the NF1 gene in NF1-specific tumours. Human Genetics, 2000, 107, 33-39.	3.8	30
222	Exploring the potential relevance of human-specific genes to complex disease. Human Genomics, 2011, 5, 99.	2.9	30
223	Interlocus gene conversion events introduce deleterious mutations into at least 1% of human genes associated with inherited disease. Genome Research, 2012, 22, 429-435.	5.5	30
224	Genome-Wide Analysis of Copy Number Variation Identifies Candidate Gene Loci Associated with the Progression of Non-Alcoholic Fatty Liver Disease. PLoS ONE, 2014, 9, e95604.	2.5	30
225	Disruption of a binding site for hepatocyte nuclear factor 1 in the protein C gene promoter is associated with hereditary thrombophilia. Human Molecular Genetics, 1994, 3, 2147-2152.	2.9	29
226	Genetic variation at the growth hormone (GH1) and growth hormone receptor (GHR) loci as a risk factor for hypertension and stroke. Human Genetics, 2006, 119, 527-540.	3.8	29
227	Genomic Medicine Without Borders: Which Strategies Should Developing Countries Employ to Invest in Precision Medicine? A New â€œFast-Second Winnerâ€•Strategy. OMICS A Journal of Integrative Biology, 2017, 21, 647-657.	2.0	29
228	Heritable pattern of oxidized DNA base repair coincides with pre-targeting of repair complexes to open chromatin. Nucleic Acids Research, 2021, 49, 221-243.	14.5	29
229	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.	6.2	29
230	Challenges in the diagnosis of neurofibromatosis type 1 (NF1) in young children facilitated by means of revised diagnostic criteria including genetic testing for pathogenic NF1 gene variants. Human Genetics, 2022, 141, 177-191.	3.8	29
231	Molecular diagnosis of facioscapulohumeral muscular dystrophy. Expert Review of Molecular Diagnostics, 2002, 2, 160-171.	3.1	28
232	Characterization of the nonallelic homologous recombination hotspot PRS3 associated with type-3NF1deletions. Human Mutation, 2012, 33, 372-383.	2.5	28
233	NAA10 polyadenylation signal variants cause syndromic microphthalmia. Journal of Medical Genetics, 2019, 56, 444-452.	3.2	28
234	Diagnosis of genetic disease using recombinant DNA. Second edition. Human Genetics, 1989, 83, 307-334.	3.8	27

#	Article	IF	Citations
235	Molecular characterisation of the pericentric inversion that distinguishes human chromosome 5 from the homologous chimpanzee chromosome. Human Genetics, 2005, 117, 168-176.	3.8	27
236	Impact of human pathogenic micro-insertions and micro-deletions on post-transcriptional regulation. Human Molecular Genetics, 2014, 23, 3024-3034.	2.9	27
237	regSNPs-splicing: a tool for prioritizing synonymous single-nucleotide substitution. Human Genetics, 2017, 136, 1279-1289.	3.8	27
238	Regulatory Mutations and Human Genetic Disease. Annals of Medicine, 1992, 24, 427-437.	3.8	26
239	p53 mutations, benzo[a]pyrene and lung cancer. Mutagenesis, 1998, 13, 319-320.	2.6	26
240	Identification and molecular characterization of six novel mutations in the UDP-N-acetylglucosamine-1-phosphotransferase gamma subunit (GNPTG) gene in patients with mucolipidosis III gamma. Human Mutation, 2009, 30, 978-984.	2.5	26
241	Identification of recurrent type-2NF1microdeletions reveals a mitotic nonallelic homologous recombination hotspot underlying a human genomic disorder. Human Mutation, 2012, 33, 1599-1609.	2.5	26
242	Application of Economic Evaluation to Assess Feasibility for Reimbursement of Genomic Testing as Part of Personalized Medicine Interventions. Frontiers in Pharmacology, 2019, 10, 830.	3.5	26
243	Population differences in the frequency of the factor V Leiden variant among people with clinically symptomatic protein C deficiency.. Journal of Medical Genetics, 1995, 32, 543-545.	3.2	25
244	Co-inheritance of a novel deletion of the entire SPINK1 gene with a CFTR missense mutation (L997F) in a family with chronic pancreatitis. Molecular Genetics and Metabolism, 2007, 92, 168-175.	1.1	25
245	Exploring the somatic NF1 mutational spectrum associated with NF1 cutaneous neurofibromas. European Journal of Human Genetics, 2012, 20, 411-419.	2.8	25
246	Transient hypermutability, chromothripsis and replication-based mechanisms in the generation of concurrent clustered mutations. Mutation Research - Reviews in Mutation Research, 2012, 750, 52-59.	5.5	25
247	Characterization of 26 deletion CNVs reveals the frequent occurrence of micro-mutations within the breakpoint-flanking regions and frequent repair of double-strand breaks by templated insertions derived from remote genomic regions. Human Genetics, 2015, 134, 589-603.	3.8	25
248	Phenotypic and genotypic overlap between mosaic NF2 and schwannomatosis in patients with multiple non-intradermal schwannomas. Human Genetics, 2018, 137, 543-552.	3.8	25
249	First estimate of the scale of canonical 5â€² splice site GT>GC variants capable of generating wildâ€type transcripts. Human Mutation, 2019, 40, 1856-1873.	2.5	25
250	Three different pathological lesions in the NF1 gene originating de novo in a family with neurofibromatosis type 1. Human Genetics, 2003, 112, 12-17.	3.8	24
251	Gross rearrangement breakpoint database (GRaBD?). Human Mutation, 2004, 23, 219-221.	2.5	24
252	Severe infantile isolated exocrine pancreatic insufficiency caused by the complete functional loss of theSPINK1gene. Human Mutation, 2017, 38, 1660-1665.	2.5	24

#	Article	IF	Citations
253	Improving the in silico assessment of pathogenicity for compensated variants. European Journal of Human Genetics, 2017, 25, 2-7.	2.8	24
254	AVADA: toward automated pathogenic variant evidence retrieval directly from the full-text literature. Genetics in Medicine, 2020, 22, 362-370.	2.4	24
255	Single base-pair substitutions in pathology and evolution: Two sides to the same coin. Human Mutation, 1996, 8, 23-31.	2.5	23
256	Origin of the prevalentSFTPBindel g.1549Câ€‰›â€‰GAA (121ins2) mutation causing surfactant protein B (SP-B) deficiency. American Journal of Medical Genetics, Part A, 2006, 140A, 62-69.	1.2	23
257	Tissue-specific differences in the proportion of mosaic large NF1 deletions are suggestive of a selective growth advantage of hematopoietic del(+/â˜) stem cells. Human Mutation, 2012, 33, 541-550.	2.5	23
258	Integrating nextâ€generation sequencing into the diagnostic testing of inherited cancer predisposition. Clinical Genetics, 2013, 83, 2-6.	2.0	23
259	Clinical heterogeneity of mitochondrial NAD kinase deficiency caused by a NADK2 start loss variant. American Journal of Medical Genetics, Part A, 2018, 176, 692-698.	1.2	23
260	Copy number variations in the NF1 gene region are infrequent and do not predispose to recurrent type-1 deletions. European Journal of Human Genetics, 2008, 16, 572-580.	2.8	22
261	Elucidation of the complex structure and origin of the human trypsinogen locus triplication. Human Molecular Genetics, 2009, 18, 3605-3614.	2.9	22
262	Revealing the human mutome. Clinical Genetics, 2010, 78, 310-320.	2.0	22
263	Bridging genomics research between developed and developing countries: the Genomic Medicine Alliance. Personalized Medicine, 2014, 11, 615-623.	1.5	22
264	A Role for Non-B DNA Forming Sequences in Mediating Microlesions Causing Human Inherited Disease. Human Mutation, 2016, 37, 65-73.	2.5	22
265	Prediction of functional regulatory SNPs in monogenic and complex disease. Human Mutation, 2011, 32, 1183-1190.	2.5	21
266	Assessment of the potential pathogenicity of missense mutations identified in the GTPase-activating protein (GAP)-related domain of the neurofibromatosis type-1 (NF1) gene. Human Mutation, 2012, 33, 1687-1696.	2.5	21
267	Molecular heterogeneity in malignant peripheral nerve sheath tumors associated with neurofibromatosis type 1. Human Genomics, 2012, 6, 18.	2.9	21
268	Genomic variants in the FTO gene are associated with sporadic amyotrophic lateral sclerosis in Greek patients. Human Genomics, 2017, 11, 30.	2.9	21
269	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.	6.2	21
270	Determinants of the factor IX mutational spectrum in haemophilia B: an analysis of missense mutations using a multi-domain molecular model of the activated protein. Human Genetics, 1994, 94, 594-608.	3.8	20

#	Article	IF	Citations
271	Identification of an intronic regulatory element in the human protein C (PROC) gene. Human Genetics, 2000, 107, 458-465.	3.8	20
272	The chimpanzee-specific pericentric inversions that distinguish humans and chimpanzees have identical breakpoints in Pan troglodytes and Pan paniscus. Genomics, 2006, 87, 39-45.	2.9	20
273	Diversity of cystathionine Î²-synthase haplotypes bearing the most common homocystinuria mutation c.833T>C: a possible role for gene conversion. Human Mutation, 2007, 28, 255-264.	2.5	20
274	Air pollution and mutations in the germline: are humans at risk?. Human Genetics, 2009, 125, 119-130.	3.8	20
275	Critical appraisal of the private genetic and pharmacogenomic testing environment in Greece. Personalized Medicine, 2011, 8, 413-420.	1.5	20
276	Ascertainment and critical assessment of the views of the general public and healthcare professionals on nutrigenomics in Greece. Personalized Medicine, 2012, 9, 201-210.	1.5	20
277	Dissecting the clinical phenotype associated with mosaic type-2 NF1 microdeletions. Neurogenetics, 2012, 13, 229-236.	1.4	20
278	A new era in the discovery of de novomutations underlying human genetic disease. Human Genomics, 2012, 6, 27.	2.9	20
279	Promoter shuffling has occurred during the evolution of the vertebrate growth hormone gene. Gene, 2000, 254, 9-18.	2.2	19
280	A novel Alu-mediated 61-kb deletion of the von Willebrand factor (VWF) gene whose breakpoints co-locate with putative matrix attachment regions. Blood Cells, Molecules, and Diseases, 2006, 36, 385-391.	1.4	19
281	No significant enrichment of rare functionally defective CPA1 variants in a large Chinese idiopathic chronic pancreatitis cohort. Human Mutation, 2017, 38, 959-963.	2.5	19
282	Neuroprotectants attenuate hypobaric hypoxia-induced brain injuries in cynomolgus monkeys. Zoological Research, 2020, 41, 3-19.	2.1	19
283	Protein C London 1: recurrent mutation at Arg 169 (CGGâ€"TGG) in the protein C gene causing thrombosis. Nucleic Acids Research, 1989, 17, 10513-10513.	14.5	18
284	Assessing the pathological relevance of SPINK1 promoter variants. European Journal of Human Genetics, 2011, 19, 1066-1073.	2.8	18
285	Elucidating Common Structural Features of Human Pathogenic Variations Using Large-Scale Atomic-Resolution Protein Networks. Human Mutation, 2014, 35, 585-593.	2.5	18
286	Local DNA dynamics shape mutational patterns of mononucleotide repeats in human genomes. Nucleic Acids Research, 2015, 43, 5065-5080.	14.5	18
287	Identification of cancer predisposition variants in apparently healthy individuals using a next-generation sequencing-based family genomics approach. Human Genomics, 2015, 9, 12.	2.9	18
288	Identification of a functional enhancer variant within the chronic pancreatitisâ€associatedSPINK1c.101A>G (p.Asn34Ser)â€containing haplotype. Human Mutation, 2017, 38, 1014-1024.	2.5	18

#	Article	IF	Citations
289	Common homozygosity for predicted loss-of-function variants reveals both redundant and advantageous effects of dispensable human genes. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 13626-13636.	7.1	18
290	Molecular Genetic Approaches to the Analysis and Diagnosis of Human Inherited Disease: An Overview. Annals of Medicine, 1992, 24, 29-42.	3.8	17
291	Polymorphic micro-inversions contribute to the genomic variability of humans and chimpanzees. Human Genetics, 2006, 119, 103-112.	3.8	17
292	Characterization of the human lineage-specific pericentric inversion that distinguishes human chromosome 1 from the homologous chromosomes of the great apes. Human Genetics, 2006, 120, 126-138.	3.8	17
293	A legal framework for biobanking: the German experience. European Journal of Human Genetics, 2007, 15, 528-532.	2.8	17
294	Enigmatic In Vivo iduronate-2-sulfatase (IDS) mutant transcript correction to wild-type in Hunter syndrome. Human Mutation, 2010, 31, E1261-E1285.	2.5	17
295	Analysis of Crossover Breakpoints Yields New Insights into the Nature of the Gene Conversion Events Associated with LargeNF1Deletions Mediated by Nonallelic Homologous Recombination. Human Mutation, 2014, 35, 215-226.	2.5	17
296	Evaluation of copy number variation and gene expression in neurofibromatosis type-1-associated malignant peripheral nerve sheath tumours. Human Genomics, 2015, 9, 3.	2.9	17
297	RegulationSpotter: annotation and interpretation of extratranscriptic DNA variants. Nucleic Acids Research, 2019, 47, W106-W113.	14.5	17
298	A platform for curated products from novel open reading frames prompts reinterpretation of disease variants. Genome Research, 2021, 31, 327-336.	5.5	17
299	The distribution of the dinucleotide CpG and cytosine methylation in the vitellogenin gene family. Journal of Molecular Evolution, 1987, 25, 107-115.	1.8	16
300	Disentangling the perturbational effects of amino acid substitutions in the DNA-binding domain of p53. Human Genetics, 1999, 104, 15-22.	3.8	16
301	Mechanism of Alu integration into the human genome. Genomic Medicine, 2007, 1, 9-17.	0.3	16
302	Growth hormone (GH1) gene variation and the growth hormone receptor (GHR) exon 3 deletion polymorphism in a West-African population. Molecular and Cellular Endocrinology, 2008, 296, 18-25.	3.2	16
303	Legal and ethical consequences of international biobanking from a national perspective: the German BMB-EUCoop project. European Journal of Human Genetics, 2010, 18, 522-525.	2.8	16
304	Gene Conversion in Human Genetic Disease. Genes, 2010, 1, 550-563.	2.4	16
305	An emerging role for microRNAs in NF1 tumorigenesis. Human Genomics, 2012, 6, 23.	2.9	16
306	Technological advances in DNA sequence enrichment and sequencing for germline genetic diagnosis. Expert Review of Molecular Diagnostics, 2012, 12, 159-173.	3.1	16

#	Article	IF	Citations
307	Clinical relevance of cancer genome sequencing. World Journal of Gastroenterology, 2013, 19, 2011.	3.3	16
308	The somatic autosomal mutation matrix in cancer genomes. Human Genetics, 2015, 134, 851-864.	3.8	16
309	The Loss and Gain of Functional Amino Acid Residues Is a Common Mechanism Causing Human Inherited Disease. PLoS Computational Biology, 2016, 12, e1005091.	3.2	16
310	The Experimentally Obtained Functional Impact Assessments of 5' Splice Site GT>GC Variants Differ Markedly from Those Predicted. Current Genomics, 2020, 21, 56-66.	1.6	16
311	Resolution of a mispaired secondary structure intermediate could account for a novel micro-insertion/deletion (387 insA/del 8 bp) in the PYGM gene causing McArdle's disease. Clinical Genetics, 2001, 59, 48-51.	2.0	15
312	Structure-based kernels for the prediction of catalytic residues and their involvement in human inherited disease. Bioinformatics, 2010, 26, 1975-1982.	4.1	15
313	A New and More Accurate Estimate of the Rate of Concurrent Tandem-Base Substitution Mutations in the Human Germline: â^¼40.4% of the Single-Nucleotide Substitution Mutation Rate. Human Mutation, 2014, 35, 392-394.	2.5	15
314	The Rise and Rise of Exome Sequencing. Public Health Genomics, 2016, 19, 315-324.	1.0	15
315	Fine mapping of meiotic NAHR-associated crossovers causing largeNF1deletions. Human Molecular Genetics, 2016, 25, 484-496.	2.9	15
316	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.	14.5	15
317	Nucleotide Weight Matrices Reveal Ubiquitous Mutational Footprints of AID/APOBEC Deaminases in Human Cancer Genomes. Cancers, 2019, 11, 211.	3.7	15
318	In silico discrimination of single nucleotide polymorphisms and pathological mutations in human gene promoter regions by means of local DNA sequence context and regularity. In Silico Biology, 2006, 6, 23-34.	0.9	15
319	A list of cloned human DNA sequences. Human Genetics, 1983, 65, 19-26.	3.8	14
320	Screening for mutations in the antithrombin III gene causing recurrent venous thrombosis by single-strand conformation polymorphism analysis. Human Mutation, 1993, 2, 324-326.	2.5	14
321	Molecular reconstruction and homology modelling of the catalytic domain of the common ancestor of the haemostatic vitamin-K-dependent serine proteinases. Human Genetics, 1996, 98, 351-370.	3.8	14
322	How to distinguish genetically between an alleged father and his monozygotic twin: A thought experiment. Forensic Science International: Genetics, 2012, 6, e129-e130.	3.1	14
323	Research and clinical applications of cancer genome sequencing. Current Opinion in Obstetrics and Gynecology, 2013, 25, 3-10.	2.0	14
324	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

#	Article	IF	Citations
325	Role of the Common PRSS1-PRSS2 Haplotype in Alcoholic and Non-Alcoholic Chronic Pancreatitis: Meta- and Re-Analyses. Genes, 2020, 11, 1349.	2.4	14
326	The Pattern of DNA Methylation in the Î´-Crystallin Genes in Transdifferentiating Neural Retina Cultures. Differentiation, 1983, 24, 33-38.	1.9	13
327	A list of cloned human DNA sequences-Supplement. Human Genetics, 1984, 67, 111-114.	3.8	13
328	The molecular genetics of familial venous thrombosis. Blood Reviews, 1991, 5, 55-70.	5.7	13
329	Single-strand conformation polymorphism (SSCP) analysis of exon 11 of the CFTR gene reliably detects more than one third of non-?F508 mutations in German cystic fibrosis patients. Human Genetics, 1992, 88, 283-7.	3.8	13
330	Hypermethylation of the neurofibromatosis type 1 (NF1) gene promoter is not a common event in the inactivation of the NF1 gene in NF1-specific tumours. Human Genetics, 2000, 107, 33-39.	3.8	13
331	Molecular genetic analysis of severe protein C deficiency. Human Genetics, 2000, 106, 646-653.	3.8	13
332	Prenatal Exclusion of Severe Factor VII Deficiency. Journal of Pediatric Hematology/Oncology, 2003, 25, 418-420.	0.6	13
333	Chromosomal speciation of humans and chimpanzees revisited: studies of DNA divergence within inverted regions. Cytogenetic and Genome Research, 2007, 116, 53-60.	1.1	13
334	A gene conversion hotspot in the human growth hormone (GH1) gene promoter. Human Mutation, 2009, 30, 239-247.	2.5	13
335	Comparative analysis of germline and somatic microlesion mutational spectra in 17 human tumor suppressor genes. Human Mutation, 2011, 32, 620-632.	2.5	13
336	regSNPs: a strategy for prioritizing regulatory single nucleotide substitutions. Bioinformatics, 2012, 28, 1879-1886.	4.1	13
337	Regulatory Single-Nucleotide Variant Predictor Increases Predictive Performance of Functional Regulatory Variants. Human Mutation, 2016, 37, 1137-1143.	2.5	13
338	Extreme clustering of type-1 NF1 deletion breakpoints co-locating with G-quadruplex forming sequences. Human Genetics, 2018, 137, 511-520.	3.8	13
339	11 The molecular genetics of familial venous thrombosis. Best Practice and Research: Clinical Haematology, 1994, 7, 637-674.	1.1	12
340	Molecular analysis of the 5â€²-flanking region of the neurofibromatosis type 1 (NF1) gene: identification of five sequence variants. Clinical Genetics, 2001, 57, 221-224.	2.0	12
341	Triangulation of the human, chimpanzee, and Neanderthal genome sequences identifies potentially compensated mutations. Human Mutation, 2010, 31, 1286-1293.	2.5	12
342	Delineating the Hemostaseome as an aid to individualize the analysis of the hereditary basis of thrombotic and bleeding disorders. Human Genetics, 2011, 130, 149-166.	3.8	12

#	Article	IF	Citations
343	Delineation of the clinical phenotype associated with non-mosaic type-2 NF1 deletions: two case reports. Journal of Medical Case Reports, 2011, 5, 577.	0.8	12
344	Restoration of the Normal Splicing Pattern of the PLP1 Gene by Means of an Antisense Oligonucleotide Directed against an Exonic Mutation. PLoS ONE, 2013, 8, e73633.	2.5	12
345	Complex Multiple-Nucleotide Substitution Mutations Causing Human Inherited Disease Reveal Novel Insights into the Action of Translesion Synthesis DNA Polymerases. Human Mutation, 2015, 36, 1034-1038.	2.5	12
346	In vitro and in silico evidence against a significant effect of theSPINK1c.194G>A variant on pre-mRNA splicing. Gut, 2017, 66, 2195-2196.	12.1	12
347	ExonImpact: Prioritizing Pathogenic Alternative Splicing Events. Human Mutation, 2017, 38, 16-24.	2.5	12
348	Pronounced maternal parent-of-origin bias for type-1 NF1 microdeletions. Human Genetics, 2018, 137, 365-373.	3.8	12
349	PRSS1 copy number variants and promoter polymorphisms in pancreatitis: common pathogenetic mechanism, different genetic effects. Gut, 2018, 67, 592-593.	12.1	12
350	Biological and functional relevance of CASP predictions. Proteins: Structure, Function and Bioinformatics, 2018, 86, 374-386.	2.6	12
351	Identification of compound heterozygous variants in the noncoding RNU4ATAC gene in a Chinese family with two successive foetuses with severe microcephaly. Human Genomics, 2018, 12, 3.	2.9	12
352	Ultra-deep amplicon sequencing indicates absence of low-grade mosaicism with normal cells in patients with type-1 NF1 deletions. Human Genetics, 2019, 138, 73-81.	3.8	12
353	Classification of NF1 microdeletions and its importance for establishing genotype/phenotype correlations in patients with NF1 microdeletions. Human Genetics, 2021, 140, 1635-1649.	3.8	12
354	Non-coding RNA ANRIL and the number of plexiform neurofibromas in patients with NF1microdeletions. BMC Medical Genetics, 2012, 13, 98.	2.1	11
355	Geneâ€“environment interaction between APOA5Âc.553G>T and pregnancy in hypertriglyceridemia-induced acute pancreatitis. Journal of Clinical Lipidology, 2020, 14, 498-506.	1.5	11
356	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.	1.2	11
357	The mutational demography of protein C deficiency. Human Genetics, 1995, 96, 142-146.	3.8	10
358	An isolated case of lissencephaly caused by the insertion of a mitochondrial genome-derived DNA sequence into the 5' untranslated region of the PAFAH1B1 (LIS1) gene. Human Genomics, 2010, 4, 384.	2.9	10
359	A Probabilistic Model to Predict Clinical Phenotypic Traits from Genome Sequencing. PLoS Computational Biology, 2014, 10, e1003825.	3.2	10
360	Genetics in Genomic Era. Genetics Research International, 2015, 2015, 1-2.	2.0	10

#	Article	IF	Citations
361	IMHOTEPâ€"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
362	Digging deeper into the intronic sequences of theSPINK1gene: TableÂ1. Gut, 2016, 65, 1055-1056.	12.1	10
363	In silico prioritization and further functional characterization of SPINK1 intronic variants. Human Genomics, 2017, 11, 7.	2.9	10
364	Analysis of the Impact of Known SPINK1 Missense Variants on Pre-mRNA Splicing and/or mRNA Stability in a Full-Length Gene Assay. Genes, 2017, 8, 263.	2.4	10
365	Verifying nomenclature of DNA variants in submitted manuscripts: Guidance for journals. Human Mutation, 2021, 42, 3-7.	2.5	10
366	Prioritization of schizophrenia risk genes from GWAS results by integrating multi-omics data. Translational Psychiatry, 2021, 11, 175.	4.8	10
367	NF1 Microdeletions and Their Underlying Mutational Mechanisms. , 2012, , 187-209.		10
368	Atypical NF1 Microdeletions: Challenges and Opportunities for Genotype/Phenotype Correlations in Patients with Large NF1 Deletions. Genes, 2021, 12, 1639.	2.4	10
369	Functional analysis of polymorphic variation within the promoter and 5? untranslated region of the neurofibromatosis type 1 (NF1) gene. American Journal of Medical Genetics Part A, 2004, 131A, 227-231.	2.4	9
370	Extended runs of homozygosity at 17q11.2: an association with type-2NF1deletions?. Human Mutation, 2010, 31, 325-334.	2.5	9
371	Small deletions within the RHD coding sequence: a report of two novel mutational events and a survey of the underlying pathophysiologic mechanisms. Transfusion, 2013, 53, 206-210.	1.6	9
372	Concurrent Nucleotide Substitution Mutations in the Human Genome Are Characterized by a Significantly Decreased Transition/Transversion Ratio. Human Mutation, 2015, 36, 333-341.	2.5	9
373	iRegNet3D: three-dimensional integrated regulatory network for the genomic analysis of coding and non-coding disease mutations. Genome Biology, 2017, 18, 10.	8.8	9
374	Scale and Scope of Gene-Alcohol Interactions in Chronic Pancreatitis: A Systematic Review. Genes, 2021, 12, 471.	2.4	9
375	Splicing Outcomes of 5â€² Splice Site GT>GC Variants That Generate Wild-Type Transcripts Differ Significantly Between Full-Length and Minigene Splicing Assays. Frontiers in Genetics, 2021, 12, 701652.	2.3	9
376	Consideration of the haplotype diversity at nonallelic homologous recombination hotspots improves the precision of rearrangement breakpoint identification. Human Mutation, 2017, 38, 1711-1722.	2.5	9
377	Human gene cloning: the storm before the lull?. Nature, 1986, 322, 119-119.	27.8	8
378	Regional localization and characterization of a DNA segment on the long arm of chromosome 21. Human Genetics, 1987, 75, 129-135.	3.8	8

#	Article	IF	Citations
379	Protein C deficiency and thromboembolism: recurrent mutation at Arg 306 in the protein C gene. Human Genetics, 1992, 88, 586-588.	3.8	8
380	Prothrombin cleavage by human vascular smooth muscle cells: A potential alternative pathway to the coagulation cascade. Journal of Cellular Biochemistry, 1995, 59, 514-528.	2.6	8
381	Core database. Nature, 1995, 374, 402-402.	27.8	8
382	Quantitative mapping of genetic similarity in human heritable diseases by shared mutations. Human Mutation, 2018, 39, 292-301.	2.5	8
383	Toward a clinical diagnostic pipeline for SPINK1 intronic variants. Human Genomics, 2019, 13, 8.	2.9	8
384	NGS mismapping confounds the clinical interpretation of the PRSS1 p.Ala16Val (c.47C>T) variant in chronic pancreatitis. Gut, 2022, 71, 841-842.	12.1	8
385	Trypsinogen (PRSS1 and PRSS2) gene dosage correlates with pancreatitis risk across genetic and transgenic studies: a systematic review and re-analysis. Human Genetics, 2022, 141, 1327-1338.	3.8	8
386	The CEL-HYB1 Hybrid Allele Promotes Digestive Enzyme Misfolding and Pancreatitis in Mice. Cellular and Molecular Gastroenterology and Hepatology, 2022, 14, 55-74.	4.5	8
387	Molecular genetic analysis of a novel form of haemophilia a characterized by the variable expression of factor VIII. Thrombosis Research, 1990, 59, 871-877.	1.7	7
388	Variation of site-specific methylation patterns in the factor VIII (F8C) gene in human sperm DNA. Human Genetics, 1998, 103, 228-233.	3.8	7
389	THE EVOLUTION OF THE VERTEBRATE ?-GLOBIN GENE PROMOTER. Evolution; International Journal of Organic Evolution, 2002, 56, 224-232.	2.3	7
390	Utilization of a cryptic noncanonical donor splice site in the KRT14 gene causes a mild form of epidermolysis bullosa simplex. British Journal of Dermatology, 2006, 155, 201-203.	1.5	7
391	Searching for potential microRNA-binding site mutations amongst known disease-associated 3â€² UTR variants. Genomic Medicine, 2007, 1, 29-33.	0.3	7
392	Exome sequencing: a transient technology for molecular diagnostics?. Expert Review of Molecular Diagnostics, 2012, 12, 211-214.	3.1	7
393	Identification of LargeNF1Duplications Reciprocal to NAHR-Mediated Type-1NF1Deletions. Human Mutation, 2014, 35, 1469-1475.	2.5	7
394	5â€² splice site GC>GT and GT>GC variants differ markedly in terms of their functionality and pathogenicity. Human Mutation, 2020, 41, 1358-1364.	2.5	7
395	Digenic Inheritance and Gene-Environment Interaction in a Patient With Hypertriglyceridemia and Acute Pancreatitis. Frontiers in Genetics, 2021, 12, 640859.	2.3	7
396	Ectopic Transcript Analysis Indicates that Allelic Exclusion is an Important Cause of Type I Protein C Deficiency in Patients with Nonsense and Frameshift Mutations in the PROC Gene. Thrombosis and Haemostasis, 1996, 75, 870-876.	3.4	7

#	Article	IF	Citations
397	Functionally deficient TRPV6 variants contribute to hereditary and familial chronic pancreatitis. Human Mutation, 2022, 43, 228-239.	2.5	7
398	A comprehensive list of cloned human DNA sequences. Nucleic Acids Research, 1990, 18, 2413-2547.	14.5	6
399	A single base-pair deletion in the protein C gene causing recurrent thromboembolism. Thrombosis Research, 1991, 61, 335-340.	1.7	6
400	The Frequency of Inherited Disorders Database. Human Genetics, 2001, 108, 72-74.	3.8	6
401	Structure-based kernels for the prediction of catalytic residues and their involvement in human inherited disease. BMC Bioinformatics, 2010, 11, .	2.6	6
402	Human Gene Mutation: Mechanisms and Consequences. , 2010, , 319-363.		6
403	The Germline Mutational Spectrum in Neurofibromatosis Type 1 and Genotypeâ€“Phenotype Correlations. , 2012, , 115-134.		6
404	Human Gene Mutation in Inherited Disease. , 2013, , 1-48.		6
405	Disclosing the Hidden Structure and Underlying Mutational Mechanism of a Novel Type of Duplication CNV Responsible for Hereditary Multiple Osteochondromas. Human Mutation, 2015, 36, 758-763.	2.5	6
406	Mining clinical attributes of genomic variants through assisted literature curation in Egas. Database: the Journal of Biological Databases and Curation, 2016, 2016, baw096.	3.0	6
407	CDG: An Online Server for Detecting Biologically Closest Disease-Causing Genes and its Application to Primary Immunodeficiency. Frontiers in Immunology, 2018, 9, 1340.	4.8	6
408	Most unambiguous loss-of-function CPA1 mutations are unlikely to predispose to chronic pancreatitis. Gut, 2020, 69, 785-786.	12.1	6
409	Pathogenic and likely pathogenic variants in at least five genes account for approximately 3% of mild isolated nonsyndromic thrombocytopenia. Transfusion, 2020, 60, 2419-2431.	1.6	6
410	The reversion variant (p.Arg90Leu) at the evolutionarily adaptive p.Arg90 site in CELA3B predisposes to chronic pancreatitis. Human Mutation, 2021, 42, 385-391.	2.5	6
411	Compensatory epistasis explored by molecular dynamics simulations. Human Genetics, 2021, 140, 1329-1342.	3.8	6
412	Common polymorphic OTC variants can act as genetic modifiers of enzymatic activity. Human Mutation, 2021, 42, 978-989.	2.5	6
413	A novel missense mutation in the antithrombin III gene (Ser349â†'Pro) causing recurrent venous thrombosis. Human Genetics, 1992, 88, 707-708.	3.8	5
414	Homology modelling of the catalytic domain of early mammalian protein C: evolution of structural features. Human Genetics, 1997, 101, 37-42.	3.8	5

#	Article	IF	Citations
415	A rare complex DNA rearrangement in the murine Steel gene results in exon duplication and a lethal phenotype. Blood, 2003, 102, 3548-3555.	1.4	5
416	Comparative analysis of copy number variation in primate genomes. Cytogenetic and Genome Research, 2008, 123, 288-296.	1.1	5
417	STREGA: a â€˜How-Toâ€™ guide for reporting genetic associations. Human Genetics, 2009, 125, 117-118.	3.8	5
418	â€˜Sifting the significance from the dataâ€™ - the impact of high-throughput genomic technologies on human genetics and health care. Human Genomics, 2012, 6, 11.	2.9	5
419	DNA structure matters. Genome Medicine, 2013, 5, 51.	8.2	5
420	Discovery and Functional Annotation ofPRSS1Promoter Variants in Chronic Pancreatitis. Human Mutation, 2016, 37, 1149-1152.	2.5	5
421	Compound Heterozygosity for Novel Truncating Variants in the LMOD3 Gene as the Cause of Polyhydramnios in Two Successive Fetuses. Frontiers in Genetics, 2019, 10, 835.	2.3	5
422	Chronic Pancreatitis: The True Pathogenic Culprit within the SPINK1 N34S-Containing Haplotype Is No Longer at Large. Genes, 2021, 12, 1683.	2.4	5
423	A comprehensive list of cloned human DNA sequences--1990 update. Nucleic Acids Research, 1991, 19, 2111-2126.	14.5	4
424	Compound heterozygosity for two novel mutations (1203insG/Y1456X) in the von Willebrand factor gene causing type 3 von Willebrand disease. Haemophilia, 2007, 13, 645-648.	2.1	4
425	Two sisters with Rett syndrome and non-identical paternally-derived microdeletions in the MECP2 gene. Genomic Medicine, 2008, 2, 77-81.	0.3	4
426	Prospects for the automated extraction of mutation data from the scientific literature. Human Genomics, 2010, 5, 1.	2.9	4
427	Is the NIH policy for sharing GWAS data running the risk of being counterproductive?. Investigative Genetics, 2010, 1, 3.	3.3	4
428	Do Inherited Disease Genes Have Distinguishing Functional Characteristics?. Genetic Testing and Molecular Biomarkers, 2010, 14, 289-291.	0.7	4
429	Lionizing lyonization 50Âyears on. Human Genetics, 2011, 130, 167-168.	3.8	4
430	Screening in silico predicted remotely acting NF1gene regulatory elements for mutations in patients with neurofibromatosis type 1. Human Genomics, 2013, 7, 18.	2.9	4
431	Intra-individual plasticity of the TAZ gene leading to different heritable mutations in siblings with Barth syndrome. European Journal of Human Genetics, 2015, 23, 1708-1712.	2.8	4
432	EXT1 and EXT2 Variants in 22 Chinese Families With Multiple Osteochondromas: Seven New Variants and Potentiation of Preimplantation Genetic Testing and Prenatal Diagnosis. Frontiers in Genetics, 2020, 11, 607838.	2.3	4

#	Article	IF	Citations
433	DNA Methylation, Deamination, and Translesion Synthesis Combine to Generate Footprint Mutations in Cancer Driver Genes in B-Cell Derived Lymphomas and Other Cancers. Frontiers in Genetics, 2021, 12, 671866.	2.3	4
434	Mutations Causing Complex Disease May under Certain Circumstances Be Protective in an Epidemiological Sense. PLoS ONE, 2015, 10, e0132150.	2.5	4
435	Molecular genetic approaches to the analysis of human ophthalmic disease. Eye, 1987, 1, 699-721.	2.1	3
436	MspI RFLP in the human heparin cofactor II (HCF2) gene. Nucleic Acids Research, 1990, 18, 1664-1664.	14.5	3
437	Detection of NF1 Mutations Utilizing the Protein Truncation Test (PTT). , 2003, 217, 315-328.		3
438	Chromosomal Distribution of Disease Genes in the Human Genome. Genetic Testing and Molecular Biomarkers, 2010, 14, 441-446.	0.7	3
439	Cross-comparison of the genome sequences from human, chimpanzee, Neanderthal and a Denisovan hominin identifies novel potentially compensated mutations. Human Genomics, 2011, 5, 453.	2.9	3
440	Local DNA sequence determinants of FUT2 copy number variation. Transfusion, 2011, 51, 1359-1361.	1.6	3
441	Analysis of Features from Protein-protein Hetero-complex Structures to Predict Protein Interaction Interfaces Using Machine Learning. Procedia Technology, 2013, 10, 62-66.	1.1	3
442	Population-specific differences in gene conversion patterns between human SUZ12 and SUZ12P are indicative of the dynamic nature of interparalog gene conversion. Human Genetics, 2014, 133, 383-401.	3.8	3
443	The sequencing and interpretation of the genome obtained from a Serbian individual. PLoS ONE, 2018, 13, e0208901.	2.5	3
444	The Somatic Mutational Spectrum of the NF1 Gene. , 2012, , 211-233.		3
445	Distinct sequence features underlie microdeletions and gross deletions in the human genome. Human Mutation, 2022, 43, 328-346.	2.5	3
446	Carrier detection in haemophilia A by direct analysis of factor VIII gene lesions. Human Genetics, 1991, 87, 99-100.	3.8	2
447	Prenatal exclusion of haemophilia a and carrier testing by direct detection of a disease lesion. Prenatal Diagnosis, 1992, 12, 861-866.	2.3	2
448	A novel missense mutation (Thr176?Ile) at the putative hinge of the neo N-terminus of activated protein C. Human Genetics, 1995, 95, 447-50.	3.8	2
449	THE EVOLUTION OF THE VERTEBRATE Î²-GLOBIN GENE PROMOTER. Evolution; International Journal of Organic Evolution, 2002, 56, 224.	2.3	2
450	Neurofibromatosis Type 1: A Common Familial Cancer Syndrome. , 2004, , 285-310.		2

#	Article	IF	Citations
451	Preface. Cytogenetic and Genome Research, 2008, 123, 5-6.	1.1	2
452	Local sequence determinants of two in-frame triplet deletion/duplication hotspots in the RHD/RHCEgenes. Human Genomics, 2012, 6, 8.	2.9	2
453	Human Genomic Variants and Inherited Disease. , 2019, , 125-200.		2
454	Sequential data selection for predicting the pathogenic effects of sequence variation. , 2015, , .		1
455	The Genomic Medicine Alliance: A Global Effort to Facilitate the Introduction of Genomics into Healthcare in Developing Nations. , 2018, , 173-188.		1
456	No Convincing Evidence to Support a Bimodal Age of Onset in Idiopathic Chronic Pancreatitis. Clinical Gastroenterology and Hepatology, 2021, , .	4.4	1
457	The Molecular Genetics of Platelet Membrane Proteins and their Inherited Disorders. Platelets, 1991, 2, 59-67.	2.3	0
458	[14]Analysis of promoter mutations causing human genetic disease. Methods in Molecular Genetics, 1996, 8, 261-277.	0.6	0
459	Molecular cytogenetic characterization of two independent karyotypic anomalies in a patient with severe mental retardation and juvenile idiopathic arthritis. Genomic Medicine, 2007, 1, 65-73.	0.3	0
460	The â€˜sequence everythingâ€™ approach and personalized clinical decision challenges. Expert Review of Molecular Diagnostics, 2012, 12, 319-322.	3.1	0
461	The Evolution of High-Throughput Sequencing Technologies: From Sanger to Single-Molecule Sequencing. , 2013, , 1-30.		0
462	Remotely acting SMCHD1 gene regulatory elements: in silico prediction and identification of potential regulatory variants in patients with FSHD. Human Genomics, 2015, 9, 25.	2.9	0
463	A Changing of the Guard at Human Genetics. Human Genetics, 2015, 134, 1-1.	3.8	0
464	In vitro recapitulation of the site-specific editing (to wild-type) of mutant IDS mRNA transcripts, and the characterization of IDS protein translated from the edited mRNAs. Human Mutation, 2017, 38, 849-862.	2.5	0
465	Somatic Copy Number Alterations: Gene and Protein Expression Correlates in NF1-Associated Malignant Peripheral Nerve Sheath Tumors. , 2012, , 405-428.		0
466	Pathogenetics of Chronic Pancreatitis. , 2017, , 63-77.		0
467	Structure and function in the human genome. , 2020, , 1-41.		0
468	Mapping the human genome. , 2020, , 43-68.		0

#	Article	IF	Citations
469	Human growth hormone I gene expression is influenced in a complex haplotype-dependent fashion by polymorphic variation in both the proximal promoter and the locus control region. Journal of Pediatric Endocrinology and Metabolism, 2002, 15 Suppl 5, 1429.	0.9	0