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	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
2	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
3	Distinct sequence features underlie microdeletions and gross deletions in the human genome. Human Mutation, 2022, 43, 328-346.	2.5	3
4	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
5	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
6	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
7	Functionally deficient TRPV6 variants contribute to hereditary and familial chronic pancreatitis. Human Mutation, 2022, 43, 228-239.	2.5	7
8	Verifying nomenclature of DNA variants in submitted manuscripts: Guidance for journals. Human Mutation, 2021, 42, 3-7.	2.5	10
9	A platform for curated products from novel open reading frames prompts reinterpretation of disease variants. Genome Research, 2021, 31, 327-336.	5.5	17
10	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
11	Scale and Scope of Gene-Alcohol Interactions in Chronic Pancreatitis: A Systematic Review. Genes, 2021, 12, 471.	2.4	9
12	Prioritization of schizophrenia risk genes from GWAS results by integrating multi-omics data. Translational Psychiatry, 2021, 11, 175.	4.8	10
13	Digenic Inheritance and Gene-Environment Interaction in a Patient With Hypertriglyceridemia and Acute Pancreatitis. Frontiers in Genetics, 2021, 12, 640859.	2.3	7
14	MutationTaster2021. Nucleic Acids Research, 2021, 49, W446-W451.	14.5	122
15	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
16	Compensatory epistasis explored by molecular dynamics simulations. Human Genetics, 2021, 140, 1329-1342.	3.8	6
17	Common polymorphic OTC variants can act as genetic modifiers of enzymatic activity. Human Mutation, 2021, 42, 978-989.	2.5	6
18	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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19	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
20	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
21	No Convincing Evidence to Support a Bimodal Age of Onset in Idiopathic Chronic Pancreatitis. Clinical Gastroenterology and Hepatology, 2021, , .	4.4	1
22	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
23	Chronic Pancreatitis: The True Pathogenic Culprit within the SPINK1 N34S-Containing Haplotype Is No Longer at Large. Genes, 2021, 12, 1683.	2.4	5
24	Atypical NF1 Microdeletions: Challenges and Opportunities for Genotype/Phenotype Correlations in Patients with Large NF1 Deletions. Genes, 2021, 12, 1639.	2.4	10
25	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
26	Most unambiguous loss-of-function CPA1 mutations are unlikely to predispose to chronic pancreatitis. Gut, 2020, 69, 785-786.	12.1	6
27	AVADA: toward automated pathogenic variant evidence retrieval directly from the full-text literature. Genetics in Medicine, 2020, 22, 362-370.	2.4	24
28	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
29	Inferring the molecular and phenotypic impact of amino acid variants with MutPred2. Nature Communications, 2020, 11, 5918.	12.8	305
30	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
31	Developmental Gene Expression Differences between Humans and Mammalian Models. Cell Reports, 2020, 33, 108308.	6.4	46
32	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
33	AMELIE speeds Mendelian diagnosis by matching patient phenotype and genotype to primary literature. Science Translational Medicine, 2020, 12, .	12.4	60
34	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
35	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
36	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

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37	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
38	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
39	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
40	Neuroprotectants attenuate hypobaric hypoxia-induced brain injuries in cynomolgus monkeys. Zoological Research, 2020, 41, 3-19.	2.1	19
41	Structure and function in the human genome. , 2020, , 1-41.		0
42	Mapping the human genome. , 2020, , 43-68.		0
43	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
44	RegulationSpotter: annotation and interpretation of extratranscriptic DNA variants. Nucleic Acids Research, 2019, 47, W106-W113.	14.5	17
45	Uganda Genome Resource Enables Insights into Population History and Genomic Discovery in Africa. Cell, 2019, 179, 984-1002.e36.	28.9	152
46	Extensive disruption of protein interactions by genetic variants across the allele frequency spectrum in human populations. Nature Communications, 2019, 10, 4141.	12.8	48
47	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
48	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
49	Gene expression across mammalian organ development. Nature, 2019, 571, 505-509.	27.8	490
50	Pathogenicity and functional impact of non-frameshifting insertion/deletion variation in the human genome. PLoS Computational Biology, 2019, 15, e1007112.	3.2	34
51	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
52	NAA10 polyadenylation signal variants cause syndromic microphthalmia. Journal of Medical Genetics, 2019, 56, 444-452.	3.2	28
53	Nucleotide Weight Matrices Reveal Ubiquitous Mutational Footprints of AID/APOBEC Deaminases in Human Cancer Genomes. Cancers, 2019, 11, 211.	3.7	15
54	S-CAP extends pathogenicity prediction to genetic variants that affect RNA splicing. Nature Genetics, 2019, 51, 755-763.	21.4	56

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55	Toward a clinical diagnostic pipeline for SPINK1 intronic variants. Human Genomics, 2019, 13, 8.	2.9	8
56	RegSNPs-intron: a computational framework for predicting pathogenic impact of intronic single nucleotide variants. Genome Biology, 2019, 20, 254.	8.8	52
57	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
58	Human Genomic Variants and Inherited Disease. , 2019, , 125-200.		2
59	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
60	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
61	Pronounced maternal parent-of-origin bias for type-1 NF1 microdeletions. Human Genetics, 2018, 137, 365-373.	3.8	12
62	PRSS1 copy number variants and promoter polymorphisms in pancreatitis: common pathogenetic mechanism, different genetic effects. Gut, 2018, 67, 592-593.	12.1	12
63	Quantitative mapping of genetic similarity in human heritable diseases by shared mutations. Human Mutation, 2018, 39, 292-301.	2.5	8
64	FATHMM-XF: accurate prediction of pathogenic point mutations via extended features. Bioinformatics, 2018, 34, 511-513.	4.1	296
65	Biological and functional relevance of CASP predictions. Proteins: Structure, Function and Bioinformatics, 2018, 86, 374-386.	2.6	12
66	DNA polymerase Î· mutational signatures are found in a variety of different types of cancer. Cell Cycle, 2018, 17, 348-355.	2.6	32
67	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
68	The sequencing and interpretation of the genome obtained from a Serbian individual. PLoS ONE, 2018, 13, e0208901.	2.5	3
69	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
70	The Genomic Medicine Alliance: A Global Effort to Facilitate the Introduction of Genomics into Healthcare in Developing Nations. , 2018, , 173-188.		1
71	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
72	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

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73	Extreme clustering of type-1 NF1 deletion breakpoints co-locating with G-quadruplex forming sequences. Human Genetics, 2018, 137, 511-520.	3.8	13
74	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
75	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
76	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
77	Emerging genotypeâ€"phenotype relationships in patients with large NF1 deletions. Human Genetics, 2017, 136, 349-376.	3.8	163
78	regSNPs-splicing: a tool for prioritizing synonymous single-nucleotide substitution. Human Genetics, 2017, 136, 1279-1289.	3.8	27
79	Mutational signatures and mutable motifs in cancer genomes. Briefings in Bioinformatics, 2017, 19, 1085-1101.	6.5	32
80	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
81	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
82	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
83	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
84	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
85	Severe infantile isolated exocrine pancreatic insufficiency caused by the complete functional loss of theSPINK1gene. Human Mutation, 2017, 38, 1660-1665.	2.5	24
86	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
87	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
88	Investigating DNA-, RNA-, and protein-based features as a means to discriminate pathogenic synonymous variants. Human Mutation, 2017, 38, 1336-1347.	2.5	37
89	In silico prioritization and further functional characterization of SPINK1 intronic variants. Human Genomics, 2017, 11, 7.	2.9	10
90	The NF1 somatic mutational landscape in sporadic human cancers. Human Genomics, 2017, 11, 13.	2.9	203

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91	ExonImpact: Prioritizing Pathogenic Alternative Splicing Events. Human Mutation, 2017, 38, 16-24.	2.5	12
92	Improving the in silico assessment of pathogenicity for compensated variants. European Journal of Human Genetics, 2017, 25, 2-7.	2.8	24
93	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
94	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
95	Genomic variants in the FTO gene are associated with sporadic amyotrophic lateral sclerosis in Greek patients. Human Genomics, 2017, 11, 30.	2.9	21
96	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
97	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
98	Pathogenetics of Chronic Pancreatitis. , 2017, , 63-77.		0
99	mutation3D: Cancer Gene Prediction Through Atomic Clustering of Coding Variants in the Structural Proteome. Human Mutation, 2016, 37, 447-456.	2.5	94
100	Test Pricing and Reimbursement in Genomic Medicine: Towards a General Strategy. Public Health Genomics, 2016, 19, 352-363.	1.0	37
101	No Association Between CELâ€“HYB Hybrid Allele and Chronic Pancreatitis in Asian Populations. Gastroenterology, 2016, 150, 1558-1560.e5.	1.3	59
102	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
103	Analysis of protein-coding genetic variation in 60,706 humans. Nature, 2016, 536, 285-291.	27.8	9,051
104	Regulatory Single-Nucleotide Variant Predictor Increases Predictive Performance of Functional Regulatory Variants. Human Mutation, 2016, 37, 1137-1143.	2.5	13
105	Discovery and Functional Annotation ofPRSS1Promoter Variants in Chronic Pancreatitis. Human Mutation, 2016, 37, 1149-1152.	2.5	5
106	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
107	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
108	The Rise and Rise of Exome Sequencing. Public Health Genomics, 2016, 19, 315-324.	1.0	15

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109	The mutation significance cutoff: gene-level thresholds for variant predictions. Nature Methods, 2016, 13, 109-110.	19.0	249
110	A Role for Non-B DNA Forming Sequences in Mediating Microlesions Causing Human Inherited Disease. Human Mutation, 2016, 37, 65-73.	2.5	22
111	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
112	Digging deeper into the intronic sequences of theSPINK1gene: TableÂ1. Gut, 2016, 65, 1055-1056.	12.1	10
113	Fine mapping of meiotic NAHR-associated crossovers causing largeNF1deletions. Human Molecular Genetics, 2016, 25, 484-496.	2.9	15
114	Clarifying the clinical relevance ofSPINK1intronic variants in chronic pancreatitis. Gut, 2016, 65, 884-886.	12.1	32
115	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
116	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
117	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
118	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
119	Genetics in Genomic Era. Genetics Research International, 2015, 2015, 1-2.	2.0	10
120	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
121	Individualized Iterative Phenotyping for Genome-wide Analysis of Loss-of-Function Mutations. American Journal of Human Genetics, 2015, 96, 913-925.	6.2	66
122	The somatic autosomal mutation matrix in cancer genomes. Human Genetics, 2015, 134, 851-864.	3.8	16
123	Sequential data selection for predicting the pathogenic effects of sequence variation. , 2015, , .		1
124	Local DNA dynamics shape mutational patterns of mononucleotide repeats in human genomes. Nucleic Acids Research, 2015, 43, 5065-5080.	14.5	18
125	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
126	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

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127	A Changing of the Guard at Human Genetics. Human Genetics, 2015, 134, 1-1.	3.8	0
128	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
129	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
130	An integrative approach to predicting the functional effects of non-coding and coding sequence variation. Bioinformatics, 2015, 31, 1536-1543.	4.1	524
131	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
132	Effect of predicted protein-truncating genetic variants on the human transcriptome. Science, 2015, 348, 666-669.	12.6	252
133	Mountain gorilla genomes reveal the impact of long-term population decline and inbreeding. Science, 2015, 348, 242-245.	12.6	326
134	A global reference for human genetic variation. Nature, 2015, 526, 68-74.	27.8	13,998
135	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
136	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
137	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
138	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
139	Mutations Causing Complex Disease May under Certain Circumstances Be Protective in an Epidemiological Sense. PLoS ONE, 2015, 10, e0132150.	2.5	4
140	Mechanisms of Base Substitution Mutagenesis in Cancer Genomes. Genes, 2014, 5, 108-146.	2.4	49
141	Key challenges for nextâ€generation pharmacogenomics. EMBO Reports, 2014, 15, 472-476.	4.5	49
142	A Probabilistic Model to Predict Clinical Phenotypic Traits from Genome Sequencing. PLoS Computational Biology, 2014, 10, e1003825.	3.2	10
143	A Massively Parallel Pipeline to Clone DNA Variants and Examine Molecular Phenotypes of Human Disease Mutations. PLoS Genetics, 2014, 10, e1004819.	3.5	47
144	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

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145	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
146	MutPred Splice: machine learning-based prediction of exonic variants that disrupt splicing. Genome Biology, 2014, 15, R19.	9.6	135
147	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
148	MutationTaster2: mutation prediction for the deep-sequencing age. Nature Methods, 2014, 11, 361-362.	19.0	3,203
149	Impact of human pathogenic micro-insertions and micro-deletions on post-transcriptional regulation. Human Molecular Genetics, 2014, 23, 3024-3034.	2.9	27
150	Elucidating Common Structural Features of Human Pathogenic Variations Using Large-Scale Atomic-Resolution Protein Networks. Human Mutation, 2014, 35, 585-593.	2.5	18
151	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
152	Bridging genomics research between developed and developing countries: the Genomic Medicine Alliance. Personalized Medicine, 2014, 11, 615-623.	1.5	22
153	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
154	Diagnostic Exome Sequencing to Elucidate the Genetic Basis of Likely Recessive Disorders in Consanguineous Families. Human Mutation, 2014, 35, 1203-1210.	2.5	75
155	Ranking non-synonymous single nucleotide polymorphisms based on disease concepts. Human Genomics, 2014, 8, 11.	2.9	163
156	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
157	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
158	Deciphering next-generation pharmacogenomics: an information technology perspective. Open Biology, 2014, 4, 140071.	3.6	41
159	Identification of LargeNF1Duplications Reciprocal to NAHR-Mediated Type-1NF1Deletions. Human Mutation, 2014, 35, 1469-1475.	2.5	7
160	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
161	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
162	New clinical and molecular insights on Barth syndrome. Orphanet Journal of Rare Diseases, 2013, 8, 27.	2.7	35

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163	DDIG-in: discriminating between disease-associated and neutral non-frameshifting micro-indels. Genome Biology, 2013, 14, R23.	9.6	63
164	Identifying Mendelian disease genes with the Variant Effect Scoring Tool. BMC Genomics, 2013, 14, S3.	2.8	360
165	Human Gene Mutation in Inherited Disease. , 2013, , 1-48.		6
166	MuPIT interactive: webserver for mapping variant positions to annotated, interactive 3D structures. Human Genetics, 2013, 132, 1235-1243.	3.8	68
167	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
168	The Evolution of High-Throughput Sequencing Technologies: From Sanger to Single-Molecule Sequencing. , 2013, , 1-30.		0
169	Genetic tests obtainable through pharmacies: the good, the bad, and the ugly. Human Genomics, 2013, 7, 17.	2.9	44
170	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
171	Interpreting Secondary Cardiac Disease Variants in an Exome Cohort. Circulation: Cardiovascular Genetics, 2013, 6, 337-346.	5.1	70
172	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
173	DNA structure matters. Genome Medicine, 2013, 5, 51.	8.2	5
174	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
175	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
176	Integrating nextâ€generation sequencing into the diagnostic testing of inherited cancer predisposition. Clinical Genetics, 2013, 83, 2-6.	2.0	23
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