David N Cooper

List of Publications by Citations

477 papers	63,491 citations	91 h-index	248 g-index
523 ext. papers	75,694 ext. citations	8.4 avg, IF	7.42 L-index

#	Paper	IF	Citations
477	A global reference for human genetic variation. *Nature*, **2015**, 526, 68-74	50.4	8599
476	Analysis of protein-coding genetic variation in 60,706 humans. *Nature*, **2016**, 536, 285-91	50.4	6940
475	A map of human genome variation from population-scale sequencing. *Nature*, **2010**, 467, 1061-73	50.4	6142
474	An integrated map of genetic variation from 1,092 human genomes. *Nature*, **2012**, 491, 56-65	50.4	6049
473	MutationTaster2: mutation prediction for the deep-sequencing age. *Nature Methods*, **2014**, 11, 361-2	21.6	2455
472	Genome sequence of the Brown Norway rat yields insights into mammalian evolution. *Nature*, **2004**, 428, 493-521	50.4	1689
471	Human Gene Mutation Database (HGMD): 2003 update. *Human Mutation*, **2003**, 21, 577-81	4.7	1206
470	Evolutionary and biomedical insights from the rhesus macaque genome. *Science*, **2007**, 316, 222-34	33.3	1072
469	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	6.3	975
468	The mutational spectrum of single base-pair substitutions in mRNA splice junctions of human genes: causes and consequences. *Human Genetics*, **1992**, 90, 41-54	6.3	946
467	A systematic survey of loss-of-function variants in human protein-coding genes. *Science*, **2012**, 335, 823-8	33.3	880
466	The CpG dinucleotide and human genetic disease. *Human Genetics*, **1988**, 78, 151-5	6.3	837
465	Mapping copy number variation by population-scale genome sequencing. *Nature*, **2011**, 470, 59-65	50.4	833
464	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	6.3	775
463	Predicting the functional, molecular, and phenotypic consequences of amino acid substitutions using hidden Markov models. *Human Mutation*, **2013**, 34, 57-65	4.7	723
462	The Human Gene Mutation Database: 2008 update. *Genome Medicine*, **2009**, 1, 13	14.4	647
461	Automated inference of molecular mechanisms of disease from amino acid substitutions. *Bioinformatics*, **2009**, 25, 2744-50	7.2	580

460	Insights into hominid evolution from the gorilla genome sequence. *Nature*, **2012**, 483, 169-75	50.4	517
459	The yak genome and adaptation to life at high altitude. *Nature Genetics*, **2012**, 44, 946-9	36.3	472
458	Gene conversion: mechanisms, evolution and human disease. *Nature Reviews Genetics*, **2007**, 8, 762-75	30.1	469
457	Demographic history and rare allele sharing among human populations. *Proceedings of the National Academy of Sciences of the United States of America*, **2011**, 108, 11983-8	11.5	455
456	M-CAP eliminates a majority of variants of uncertain significance in clinical exomes at high sensitivity. *Nature Genetics*, **2016**, 48, 1581-1586	36.3	423
455	Variation in genome-wide mutation rates within and between human families. *Nature Genetics*, **2011**, 43, 712-4	36.3	404
454	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, 1077-130	6.3	384
453	Gene deletions causing human genetic disease: mechanisms of mutagenesis and the role of the local DNA sequence environment. *Human Genetics*, **1991**, 86, 425-41	6.3	357
452	An integrative approach to predicting the functional effects of non-coding and coding sequence variation. *Bioinformatics*, **2015**, 31, 1536-43	7.2	340
451	The mutational spectrum of single base-pair substitutions causing human genetic disease: patterns and predictions. *Human Genetics*, **1990**, 85, 55-74	6.3	304
450	Single base-pair substitutions in exon-intron junctions of human genes: nature, distribution, and consequences for mRNA splicing. *Human Mutation*, **2007**, 28, 150-8	4.7	277
449	Cytosine methylation and the fate of CpG dinucleotides in vertebrate genomes. *Human Genetics*, **1989**, 83, 181-8	6.3	269
448	An estimate of unique DNA sequence heterozygosity in the human genome. *Human Genetics*, **1985**, 69, 201-5	6.3	269
447	Neighboring-nucleotide effects on the rates of germ-line single-base-pair substitution in human genes. *American Journal of Human Genetics*, **1998**, 63, 474-88	11	256
446	Identifying Mendelian disease genes with the variant effect scoring tool. *BMC Genomics*, **2013**, 14 Suppl 3, S3	4.5	240
445	A meta-analysis of nonsense mutations causing human genetic disease. *Human Mutation*, **2008**, 29, 1037-47	4.7	238
444	Splicing factor SFRS1 recognizes a functionally diverse landscape of RNA transcripts. *Genome Research*, **2009**, 19, 381-94	9.7	230
443	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, 1022-32	11	221

442	Genome sequencing and comparison of two nonhuman primate animal models, the cynomolgus and Chinese rhesus macaques. *Nature Biotechnology*, **2011**, 29, 1019-23	44.5	219
441	The human gene mutation database. *Trends in Genetics*, **1997**, 13, 121-2	8.5	219
440	Translocation and gross deletion breakpoints in human inherited disease and cancer I: Nucleotide composition and recombination-associated motifs. *Human Mutation*, **2003**, 22, 229-44	4.7	203
439	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, 1119-29	5.6	196
438	Mountain gorilla genomes reveal the impact of long-term population decline and inbreeding. *Science*, **2015**, 348, 242-245	33.3	195
437	Human gene mutation database-a biomedical information and research resource. *Human Mutation*, **2000**, 15, 45-51	4.7	187
436	Gene expression across mammalian organ development. *Nature*, **2019**, 571, 505-509	50.4	179
435	The evaluation of tools used to predict the impact of missense variants is hindered by two types of circularity. *Human Mutation*, **2015**, 36, 513-23	4.7	178
434	Eukaryotic DNA methylation. *Human Genetics*, **1983**, 64, 315-33	6.3	178
433	A systematic analysis of LINE-1 endonuclease-dependent retrotranspositional events causing human genetic disease. *Human Genetics*, **2005**, 117, 411-27	6.3	173
432	The mutation significance cutoff: gene-level thresholds for variant predictions. *Nature Methods*, **2016**, 13, 109-10	21.6	171
431	Human genomics. Effect of predicted protein-truncating genetic variants on the human transcriptome. *Science*, **2015**, 348, 666-9	33.3	170
430	Gains of glycosylation comprise an unexpectedly large group of pathogenic mutations. *Nature Genetics*, **2005**, 37, 692-700	36.3	168
429	The Human Gene Mutation Database (HGMD) and its exploitation in the fields of personalized genomics and molecular evolution. *Current Protocols in Bioinformatics*, **2012**, Chapter 1, Unit1.13	24.2	162
428	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, 14162-7	11.5	162
427	Unmethylated domains in vertebrate DNA. *Nucleic Acids Research*, **1983**, 11, 647-58	20.1	162
426	The functional spectrum of low-frequency coding variation. *Genome Biology*, **2011**, 12, R84	18.3	161
425	Predicting the functional consequences of cancer-associated amino acid substitutions. *Bioinformatics*, **2013**, 29, 1504-10	7.2	154

424	The human gene mutation database. *Nucleic Acids Research*, **1998**, 26, 285-7	20.1	153
423	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, 13615-20	11.5	152
422	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, 591-7	6.3	151
421	Functional intronic polymorphisms: Buried treasure awaiting discovery within our genes. *Human Genomics*, **2010**, 4, 284-8	6.8	149
420	FATHMM-XF: accurate prediction of pathogenic point mutations via extended features. *Bioinformatics*, **2018**, 34, 511-513	7.2	147
419	Early onset seizures and Rett-like features associated with mutations in CDKL5. *European Journal of Human Genetics*, **2005**, 13, 1113-20	5.3	143
418	DNA restriction fragment length polymorphisms and heterozygosity in the human genome. *Human Genetics*, **1984**, 66, 1-16	6.3	143
417	Genes, mutations, and human inherited disease at the dawn of the age of personalized genomics. *Human Mutation*, **2010**, 31, 631-55	4.7	138
416	The molecular genetics of growth hormone deficiency. *Human Genetics*, **1998**, 103, 255-72	6.3	135
415	A systematic analysis of disease-associated variants in the 3' regulatory regions of human protein-coding genes I: general principles and overview. *Human Genetics*, **2006**, 120, 1-21	6.3	131
414	The NF1 somatic mutational landscape in sporadic human cancers. *Human Genomics*, **2017**, 11, 13	6.8	130
413	Clinical characterisation of 29 neurofibromatosis type-1 patients with molecularly ascertained 1.4 Mb type-1 NF1 deletions. *Journal of Medical Genetics*, **2010**, 47, 623-30	5.8	128
412	Meta-analysis of gross insertions causing human genetic disease: novel mutational mechanisms and the role of replication slippage. *Human Mutation*, **2005**, 25, 207-21	4.7	128
411	Microdeletions and microinsertions causing human genetic disease: common mechanisms of mutagenesis and the role of local DNA sequence complexity. *Human Mutation*, **2005**, 26, 205-13	4.7	128
410	Exome sequencing: dual role as a discovery and diagnostic tool. *Annals of Neurology*, **2012**, 71, 5-14	9.4	126
409	Emerging genotype-phenotype relationships in patients with large NF1 deletions. *Human Genetics*, **2017**, 136, 349-376	6.3	125
408	Loss of exon identity is a common mechanism of human inherited disease. *Genome Research*, **2011**, 21, 1563-71	9.7	125
407	Precursor-product relationship between vitellogenin and the yolk proteins as derived from the complete sequence of a Xenopus vitellogenin gene. *Nucleic Acids Research*, **1987**, 15, 4737-60	20.1	122

406	A systematic analysis of disease-associated variants in the 3' regulatory regions of human protein-coding genes II: the importance of mRNA secondary structure in assessing the functionality of 3' UTR variants. *Human Genetics*, **2006**, 120, 301-33	6.3	119
405	Genomic rearrangements in inherited disease and cancer. *Seminars in Cancer Biology*, **2010**, 20, 222-33	12.7	118
404	The Human Gene Mutation Database (HGMD): optimizing its use in a clinical diagnostic or research setting. *Human Genetics*, **2020**, 139, 1197-1207	6.3	108
403	Human gene mutations affecting RNA processing and translation. *Annals of Medicine*, **1993**, 25, 11-7	1.5	108
402	Ranking non-synonymous single nucleotide polymorphisms based on disease concepts. *Human Genomics*, **2014**, 8, 11	6.8	105
401	MutPred Splice: machine learning-based prediction of exonic variants that disrupt splicing. *Genome Biology*, **2014**, 15, R19	18.3	102
400	Evolutionary conservation and selection of human disease gene orthologs in the rat and mouse genomes. *Genome Biology*, **2004**, 5, R47	18.3	102
399	Genomic rearrangements in the CFTR gene: extensive allelic heterogeneity and diverse mutational mechanisms. *Human Mutation*, **2004**, 23, 343-57	4.7	101
398	CRAVAT: cancer-related analysis of variants toolkit. *Bioinformatics*, **2013**, 29, 647-8	7.2	98
397	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	4.7	98
396	Human genetic disease caused by de novo mitochondrial-nuclear DNA transfer. *Human Genetics*, **2003**, 112, 303-9	6.3	96
395	Neurofibromatosis type 1-associated tumours: their somatic mutational spectrum and pathogenesis. *Human Genomics*, **2011**, 5, 623-90	6.8	94
394	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, 406-10	6.8	94
393	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, 408-23	4.7	93
392	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, 424-40	4.7	91
391	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, 245-51	4.7	91
390	Mechanisms of insertional mutagenesis in human genes causing genetic disease. *Human Genetics*, **1991**, 87, 409-15	6.3	91
389	DNA polymorphism and the study of disease associations. *Human Genetics*, **1988**, 78, 299-312	6.3	91

388	Mosaic type-1 NF1 microdeletions as a cause of both generalized and segmental neurofibromatosis type-1 (NF1). *Human Mutation*, **2011**, 32, 213-9	4.7	89
387	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, 134-46	4.7	89
386	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, 1075-99	4.7	88
385	Complex gene rearrangements caused by serial replication slippage. *Human Mutation*, **2005**, 26, 125-34	4.7	85
384	Inferring the molecular and phenotypic impact of amino acid variants with MutPred2. *Nature Communications*, **2020**, 11, 5918	17.4	84
383	Analysis of protein-coding genetic variation in 60,706 humans		81
382	Abundance and length of simple repeats in vertebrate genomes are determined by their structural properties. *Genome Research*, **2008**, 18, 1545-53	9.7	79
381	Haemophilia A: database of nucleotide substitutions, deletions, insertions and rearrangements of the factor VIII gene, second edition. *Nucleic Acids Research*, **1994**, 22, 3511-33	20.1	79
380	MutPred2: inferring the molecular and phenotypic impact of amino acid variants		79
379	Understanding the recent evolution of the human genome: insights from human-chimpanzee genome comparisons. *Human Mutation*, **2007**, 28, 99-130	4.7	78
378	Human Gene Mutation Database: towards a comprehensive central mutation database. *Journal of Medical Genetics*, **2008**, 45, 124-6	5.8	77
377	Translocation and deletion breakpoints in cancer genomes are associated with potential non-B DNA-forming sequences. *Nucleic Acids Research*, **2016**, 44, 5673-88	20.1	77
376	Uganda Genome Resource Enables Insights into Population History and Genomic Discovery in Africa. *Cell*, **2019**, 179, 984-1002.e36	56.2	76
375	Identification and characterization of 15 novel GALC gene mutations causing Krabbe disease. *Human Mutation*, **2010**, 31, E1894-914	4.7	74
374	Mutational and functional analysis of the neurofibromatosis type 1 (NF1) gene. *Human Genetics*, **1997**, 99, 88-92	6.3	73
373	Human type I hair keratin pseudogene phihHaA 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	6.3	71
372	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, 567-76	5.3	70
371	The molecular pathogenesis of schwannomatosis, a paradigm for the co-involvement of multiple tumour suppressor genes in tumorigenesis. *Human Genetics*, **2017**, 136, 129-148	6.3	69

370	Report of the DNA committee and catalogues of cloned and mapped genes, markers formatted for PCR and DNA polymorphisms. *Cytogenetic and Genome Research*, **1991**, 58, 1190-1832	1.9	69
369	A new paradigm emerges from the study of de novo mutations in the context of neurodevelopmental disease. *Molecular Psychiatry*, **2013**, 18, 141-53	15.1	68
368	Restriction fragment length polymorphisms at the human parathyroid hormone gene locus. *Human Genetics*, **1984**, 67, 428-31	6.3	68
367	Inherited Factor VII Deficiency: Molecular Genetics and Pathophysiology. *Thrombosis and Haemostasis*, **1997**, 78, 151-160	7	68
366	Inherited Factor X Deficiency: Molecular Genetics and Pathophysiology. *Thrombosis and Haemostasis*, **1997**, 78, 161-172	7	68
365	Diagnostic exome sequencing to elucidate the genetic basis of likely recessive disorders in consanguineous families. *Human Mutation*, **2014**, 35, 1203-10	4.7	67
364	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	3.7	66
363	Assessing the Pathogenicity of Insertion and Deletion Variants with the Variant Effect Scoring Tool (VEST-Indel). *Human Mutation*, **2016**, 37, 28-35	4.7	65
362	Human genetics and genomics a decade after the release of the draft sequence of the human genome. *Human Genomics*, **2011**, 5, 577-622	6.8	65
361	Evaluation of denaturing high performance liquid chromatography (DHPLC) for the mutational analysis of the neurofibromatosis type 1 (NF1) gene. *Human Genetics*, **2001**, 109, 487-97	6.3	64
360	GWAS: heritability missing in action?. *European Journal of Human Genetics*, **2010**, 18, 859-61	5.3	62
359	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, 270-84	6.3	62
358	Diagnosis of genetic disease using recombinant DNA. *Human Genetics*, **1986**, 73, 1-11	6.3	61
357	Mosaicism in sporadic neurofibromatosis type 1: variations on a theme common to other hereditary cancer syndromes?. *Journal of Medical Genetics*, **2008**, 45, 622-31	5.8	60
356	Interpreting secondary cardiac disease variants in an exome cohort. *Circulation: Cardiovascular Genetics*, **2013**, 6, 337-46		59
355	Molecular mechanisms of chromosomal rearrangement during primate evolution. *Chromosome Research*, **2008**, 16, 41-56	4.4	59
354	Gonosomal mosaicism for a nonsense mutation (R1947X) in the NF1 gene in segmental neurofibromatosis type 1. *Journal of Investigative Dermatology*, **2005**, 125, 463-6	4.3	59
353	Intrachromosomal serial replication slippage in trans gives rise to diverse genomic rearrangements involving inversions. *Human Mutation*, **2005**, 26, 362-73	4.7	58

352	Gross rearrangements of the MECP2 gene are found in both classical and atypical Rett syndrome patients. *Journal of Medical Genetics*, **2006**, 43, 451-6	5.8	58
351	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, 123-8	6.3	58
350	Gain-of-glycosylation mutations. *Current Opinion in Genetics and Development*, **2007**, 17, 245-51	4.9	57
349	MuPIT interactive: webserver for mapping variant positions to annotated, interactive 3D structures. *Human Genetics*, **2013**, 132, 1235-43	6.3	56
348	IDUA mutational profiling of a cohort of 102 European patients with mucopolysaccharidosis type I: identification and characterization of 35 novel α-L-iduronidase (IDUA) alleles. *Human Mutation*, **2011**, 32, E2189-210	4.7	56
347	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, 1189-98	4.7	56
346	In silico functional profiling of human disease-associated and polymorphic amino acid substitutions. *Human Mutation*, **2010**, 31, 335-46	4.7	55
345	Microattribution and nanopublication as means to incentivize the placement of human genome variation data into the public domain. *Human Mutation*, **2012**, 33, 1503-12	4.7	54
344	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, 1201-20	11	54
343	Using exome data to identify malignant hyperthermia susceptibility mutations. *Anesthesiology*, **2013**, 119, 1043-53	4.3	53
342	DDIG-in: discriminating between disease-associated and neutral non-frameshifting micro-indels. *Genome Biology*, **2013**, 14, R23	18.3	52
341	Long homopurine*homopyrimidine sequences are characteristic of genes expressed in brain and the pseudoautosomal region. *Nucleic Acids Research*, **2006**, 34, 2663-75	20.1	52
340	Haemophilia A: database of nucleotide substitutions, deletions, insertions and rearrangements of the factor VIII gene, second edition. *Nucleic Acids Research*, **1994**, 22, 4851-68	20.1	52
339	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	4.7	52
338	mutation3D: Cancer Gene Prediction Through Atomic Clustering of Coding Variants in the Structural Proteome. *Human Mutation*, **2016**, 37, 447-56	4.7	51
337	Disease-causing mutations in the human genome. *European Journal of Pediatrics*, **2000**, 159 Suppl 3, S173-8	4.8	50
336	Estimating the efficacy and efficiency of cascade genetic screening. *American Journal of Human Genetics*, **2001**, 69, 361-70	11	49
335	DNA methylation and CpG suppression. *Cell Differentiation*, **1985**, 17, 199-205		48

334	SPINK1, PRSS1, CTRC, and CFTR Genotypes Influence Disease Onset and Clinical Outcomes in Chronic Pancreatitis. *Clinical and Translational Gastroenterology*, **2018**, 9, 204	4.2	48
333	Individualized iterative phenotyping for genome-wide analysis of loss-of-function mutations. *American Journal of Human Genetics*, **2015**, 96, 913-25	11	47
332	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	46
331	Closely spaced multiple mutations as potential signatures of transient hypermutability in human genes. *Human Mutation*, **2009**, 30, 1435-48	4.7	45
330	Report of the DNA committee and catalogues of cloned and mapped genes and DNA polymorphisms. *Cytogenetic and Genome Research*, **1990**, 55, 457-778	1.9	45
329	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, 1599-606	7.2	44
328	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	18.3	44
327	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	4.7	44
326	Molecular analysis of the genotype-phenotype relationship in factor X deficiency. *Human Genetics*, **2000**, 106, 249-57	6.3	44
325	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, 1068-75	5.6	43
324	The effect of replication errors on the mismatch analysis of PCR-amplified DNA. *Nucleic Acids Research*, **1990**, 18, 973-8	20.1	43
323	The Human Gene Mutation Database (HGMD) and its exploitation in the study of mutational mechanisms. *Current Protocols in Bioinformatics*, **2006**, Chapter 1, Unit 1.13	24.2	42
322	Key challenges for next-generation pharmacogenomics: Science & Society series on Science and Drugs. *EMBO Reports*, **2014**, 15, 472-6	6.5	41
321	Genotype-phenotype associations in neurofibromatosis type 1 (NF1): an increased risk of tumor complications in patients with NF1 splice-site mutations?. *Human Genomics*, **2012**, 6, 12	6.8	41
320	A critical view of the general public's awareness and physicians' opinion of the trends and potential pitfalls of genetic testing in Greece. *Personalized Medicine*, **2011**, 8, 551-561	2.2	41
319	Characterisation of a functional intronic polymorphism in the human growth hormone (GH1) gene. *Human Genomics*, **2010**, 4, 289-301	6.8	41
318	Structural divergence between the human and chimpanzee genomes. *Human Genetics*, **2007**, 120, 759-78	6.3	41
317	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, 763-76	4.7	40

316	Assessing radiation-associated mutational risk to the germline: repetitive DNA sequences as mutational targets and biomarkers. *Radiation Research*, **2006**, 165, 249-68	3.1	40
315	No Association Between CEL-HYB Hybrid Allele and Chronic Pancreatitis in Asian Populations. *Gastroenterology*, **2016**, 150, 1558-1560.e5	13.3	40
314	Genetic tests obtainable through pharmacies: the good, the bad, and the ugly. *Human Genomics*, **2013**, 7, 17	6.8	38
313	Critical appraisal of the views of healthcare professionals with respect to pharmacogenomics and personalized medicine in Greece. *Personalized Medicine*, **2014**, 11, 15-26	2.2	38
312	Independent intrachromosomal recombination events underlie the pericentric inversions of chimpanzee and gorilla chromosomes homologous to human chromosome 16. *Genome Research*, **2005**, 15, 1232-42	9.7	38
311	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, 742-51	4.7	37
310	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, 56182		36
309	A massively parallel pipeline to clone DNA variants and examine molecular phenotypes of human disease mutations. *PLoS Genetics*, **2014**, 10, e1004819	6	35
308	Molecular characterization of 22 novel UDP-N-acetylglucosamine-1-phosphate transferase alpha- and beta-subunit (GNPTAB) gene mutations causing mucolipidosis types IIalpha/beta and IIIalpha/beta in 46 patients. *Human Mutation*, **2009**, 30, E956-73	4.7	35
307	Ectopic (illegitimate) transcription: new possibilities for the analysis and diagnosis of human genetic disease. *Annals of Medicine*, **1994**, 26, 9-14	1.5	35
306	Chicken lens delta-crystallin gene expression and methylation in several non-lens tissues. *Nucleic Acids Research*, **1983**, 11, 2513-27	20.1	35
305	Mechanisms of loss of heterozygosity in neurofibromatosis type 1-associated plexiform neurofibromas. *Journal of Investigative Dermatology*, **2009**, 129, 615-21	4.3	34
304	Identification of large-scale human-specific copy number differences by inter-species array comparative genomic hybridization. *Human Genetics*, **2006**, 119, 185-98	6.3	34
303	Assessing the relative importance of the biophysical properties of amino acid substitutions associated with human genetic disease. *Human Mutation*, **2002**, 20, 98-109	4.7	34
302	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	7	34
301	Deciphering next-generation pharmacogenomics: an information technology perspective. *Open Biology*, **2014**, 4,	7	33
300	Evolution of the proximal promoter region of the mammalian growth hormone gene. *Gene*, **1999**, 237, 143-51	3.8	33
299	Mechanisms of base substitution mutagenesis in cancer genomes. *Genes*, **2014**, 5, 108-46	4.2	32

298	Gene discovery in familial cancer syndromes by exome sequencing: prospects for the elucidation of familial colorectal cancer type X. *Modern Pathology*, **2012**, 25, 1055-68	9.8	31
297	Down's syndrome and the molecular biology of chromosome 21. *Progress in Neurobiology*, **1988**, 30, 507-30	10.9	31
296	Meiotic recombination favors the spreading of deleterious mutations in human populations. *Human Mutation*, **2011**, 32, 198-206	4.7	30
295	Single base-pair substitutions at the translation initiation sites of human genes as a cause of inherited disease. *Human Mutation*, **2011**, 32, 1137-43	4.7	30
294	Molecular genetic analysis of severe protein C deficiency. *Human Genetics*, **2000**, 106, 646-53	6.3	30
293	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, 203-11	5.8	29
292	Exome versus transcriptome sequencing in identifying coding region variants. *Expert Review of Molecular Diagnostics*, **2012**, 12, 241-51	3.8	29
291	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, 421-8	4.7	29
290	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, 33-9	6.3	29
289	Intrachromosomal mitotic nonallelic homologous recombination is the major molecular mechanism underlying type-2 NF1 deletions. *Human Mutation*, **2010**, 31, 1163-73	4.7	28
288	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, 1359-61	4.3	28
287	Late-onset homozygous protein C deficiency. *Lancet, The*, **1991**, 338, 575-6	40	28
286	Test Pricing and Reimbursement in Genomic Medicine: Towards a General Strategy. *Public Health Genomics*, **2016**, 19, 352-363	1.9	28
285	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	6.8	27
284	Non-B DNA-forming sequences and WRN deficiency independently increase the frequency of base substitution in human cells. *Journal of Biological Chemistry*, **2011**, 286, 10017-26	5.4	27
283	Detection of two Alu insertions in the CFTR gene. *Journal of Cystic Fibrosis*, **2008**, 7, 37-43	4.1	27
282	Human gene mutation in pathology and evolution. *Journal of Inherited Metabolic Disease*, **2002**, 25, 157-82	3.4	27
281	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, 2147-52	5.6	27

280	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	3.7	27
279	New clinical and molecular insights on Barth syndrome. *Orphanet Journal of Rare Diseases*, **2013**, 8, 27	4.2	26
278	Investigating DNA-, RNA-, and protein-based features as a means to discriminate pathogenic synonymous variants. *Human Mutation*, **2017**, 38, 1336-1347	4.7	26
277	A meta-analysis of single base-pair substitutions in translational termination codons ('nonstop' mutations) that cause human inherited disease. *Human Genomics*, **2011**, 5, 241-64	6.8	26
276	Monozygotic twins discordant for neurofibromatosis type 1 due to a postzygotic NF1 gene mutation. *Human Mutation*, **2011**, 32, E2134-47	4.7	26
275	Detection of missense mutations by single-strand conformational polymorphism (SSCP) analysis in five dysfunctional variants of coagulation factor VII. *Human Molecular Genetics*, **1993**, 2, 1355-9	5.6	26
274	Diagnosis of genetic disease using recombinant DNA. Second edition. *Human Genetics*, **1989**, 83, 307-34	6.3	26
273	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	11.5	26
272	Mutational signatures and mutable motifs in cancer genomes. *Briefings in Bioinformatics*, **2018**, 19, 1085-1101	13.4	25
271	S-CAP extends pathogenicity prediction to genetic variants that affect RNA splicing. *Nature Genetics*, **2019**, 51, 755-763	36.3	25
270	Impact of human pathogenic micro-insertions and micro-deletions on post-transcriptional regulation. *Human Molecular Genetics*, **2014**, 23, 3024-34	5.6	25
269	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	4.2	25
268	Cruciform-forming inverted repeats appear to have mediated many of the microinversions that distinguish the human and chimpanzee genomes. *Chromosome Research*, **2009**, 17, 469-83	4.4	25
267	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, 527-40	6.3	25
266	RegSNPs-intron: a computational framework for predicting pathogenic impact of intronic single nucleotide variants. *Genome Biology*, **2019**, 20, 254	18.3	25
265	Proteins linked to autosomal dominant and autosomal recessive disorders harbor characteristic rare missense mutation distribution patterns. *Human Molecular Genetics*, **2015**, 24, 5995-6002	5.6	24
264	Transient hypermutability, chromothripsis and replication-based mechanisms in the generation of concurrent clustered mutations. *Mutation Research - Reviews in Mutation Research*, **2012**, 750, 52-9	7	24
263	Guanine holes are prominent targets for mutation in cancer and inherited disease. *PLoS Genetics*, **2013**, 9, e1003816	6	24

262	Exploring the potential relevance of human-specific genes to complex disease. *Human Genomics*, **2011**, 5, 99-107	6.8	24
261	Gross Rearrangement Breakpoint Database (GRaBD). *Human Mutation*, **2004**, 23, 219-21	4.7	24
260	Extensive disruption of protein interactions by genetic variants across the allele frequency spectrum in human populations. *Nature Communications*, **2019**, 10, 4141	17.4	23
259	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, 435-58	6.3	23
258	Characterization of the nonallelic homologous recombination hotspot PRS3 associated with type-3 NF1 deletions. *Human Mutation*, **2012**, 33, 372-83	4.7	23
257	Genomic Medicine Without Borders: Which Strategies Should Developing Countries Employ to Invest in Precision Medicine? A New "Fast-Second Winner" Strategy. *OMICS A Journal of Integrative Biology*, **2017**, 21, 647-657	3.8	23
256	When loss-of-function is loss of function: assessing mutational signatures and impact of loss-of-function genetic variants. *Bioinformatics*, **2017**, 33, i389-i398	7.2	23
255	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, 978-84	4.7	23
254	Interlocus gene conversion events introduce deleterious mutations into at least 1% of human genes associated with inherited disease. *Genome Research*, **2012**, 22, 429-35	9.7	23
253	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, 168-75	3.7	23
252	p53 mutations, benzo[a]pyrene and lung cancer. *Mutagenesis*, **1998**, 13, 319-20	2.8	23
251	Regulatory mutations and human genetic disease. *Annals of Medicine*, **1992**, 24, 427-37	1.5	23
250	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	6.3	22
249	Clarifying the clinical relevance of SPINK1 intronic variants in chronic pancreatitis. *Gut*, **2016**, 65, 884-6	19.2	22
248	Patterns and mutational signatures of tandem base substitutions causing human inherited disease. *Human Mutation*, **2013**, 34, 1119-30	4.7	22
247	Exploring the somatic NF1 mutational spectrum associated with NF1 cutaneous neurofibromas. *European Journal of Human Genetics*, **2012**, 20, 411-9	5.3	22
246	Three different pathological lesions in the NF1 gene originating de novo in a family with neurofibromatosis type 1. *Human Genetics*, **2003**, 112, 12-7	6.3	22
245	Molecular characterisation of the pericentric inversion that distinguishes human chromosome 5 from the homologous chimpanzee chromosome. *Human Genetics*, **2005**, 117, 168-76	6.3	22

244	DNA polymerase mutational signatures are found in a variety of different types of cancer. *Cell Cycle*, **2018**, 17, 348-355	4.7	22
243	Bridging genomics research between developed and developing countries: the Genomic Medicine Alliance. *Personalized Medicine*, **2014**, 11, 615-623	2.2	21
242	Identification of recurrent type-2 NF1 microdeletions reveals a mitotic nonallelic homologous recombination hotspot underlying a human genomic disorder. *Human Mutation*, **2012**, 33, 1599-609	4.7	21
241	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, 1687-96	4.7	20
240	Critical appraisal of the private genetic and pharmacogenomic testing environment in Greece. *Personalized Medicine*, **2011**, 8, 413-420	2.2	20
239	Prediction of functional regulatory SNPs in monogenic and complex disease. *Human Mutation*, **2011** , 32, 1183-90	4.7	20
238	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, 572-80	5.3	20
237	Origin of the prevalent SFTPB indel g.1549C > GAA (121ins2) mutation causing surfactant protein B (SP-B) deficiency. *American Journal of Medical Genetics, Part A*, **2006**, 140, 62-9	2.5	20
236	An integrative approach to predicting the functional effects of small indels in non-coding regions of the human genome. *BMC Bioinformatics*, **2017**, 18, 442	3.6	19
235	Tissue-specific differences in the proportion of mosaic large NF1 deletions are suggestive of a selective growth advantage of hematopoietic del(+/-) stem cells. *Human Mutation*, **2012**, 33, 541-50	4.7	19
234	Integrating next-generation sequencing into the diagnostic testing of inherited cancer predisposition. *Clinical Genetics*, **2013**, 83, 2-6	4	19
233	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, 385-91	2.1	19
232	Molecular diagnosis of facioscapulohumeral muscular dystrophy. *Expert Review of Molecular Diagnostics*, **2002**, 2, 160-71	3.8	19
231	Promoter shuffling has occurred during the evolution of the vertebrate growth hormone gene. *Gene*, **2000**, 254, 9-18	3.8	19
230	Elucidation of the complex structure and origin of the human trypsinogen locus triplication. *Human Molecular Genetics*, **2009**, 18, 3605-14	5.6	18
229	Ascertainment and critical assessment of the views of the general public and healthcare professionals on nutrigenomics in Greece. *Personalized Medicine*, **2012**, 9, 201-210	2.2	18
228	Diversity of cystathionine beta-synthase haplotypes bearing the most common homocystinuria mutation c.833T>C: a possible role for gene conversion. *Human Mutation*, **2007**, 28, 255-64	4.7	18
227	Single base-pair substitutions in pathology and evolution: two sides to the same coin. *Human Mutation*, **1996**, 8, 23-31	4.7	18

226	Identification of cancer predisposition variants in apparently healthy individuals using a next-generation sequencing-based family genomics approach. *Human Genomics*, **2015**, 9, 12	6.8	17
225	Dissecting the clinical phenotype associated with mosaic type-2 NF1 microdeletions. *Neurogenetics*, **2012**, 13, 229-36	3	17
224	Revealing the human mutome. *Clinical Genetics*, **2010**, 78, 310-20	4	17
223	Air pollution and mutations in the germline: are humans at risk?. *Human Genetics*, **2009**, 125, 119-30	6.3	17
222	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, 126-38	6.3	17
221	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, 543-5	5.8	17
220	Elucidating common structural features of human pathogenic variations using large-scale atomic-resolution protein networks. *Human Mutation*, **2014**, 35, 585-93	4.7	16
219	Molecular heterogeneity in malignant peripheral nerve sheath tumors associated with neurofibromatosis type 1. *Human Genomics*, **2012**, 6, 18	6.8	16
218	A legal framework for biobanking: the German experience. *European Journal of Human Genetics*, **2007**, 15, 528-32	5.3	16
217	The chimpanzee-specific pericentric inversions that distinguish humans and chimpanzees have identical breakpoints in Pan troglodytes and Pan paniscus. *Genomics*, **2006**, 87, 39-45	4.3	16
216	Identification of an intronic regulatory element in the human protein C (PROC) gene. *Human Genetics*, **2000**, 107, 458-65	6.3	16
215	Heritable pattern of oxidized DNA base repair coincides with pre-targeting of repair complexes to open chromatin. *Nucleic Acids Research*, **2021**, 49, 221-243	20.1	16
214	regSNPs-splicing: a tool for prioritizing synonymous single-nucleotide substitution. *Human Genetics*, **2017**, 136, 1279-1289	6.3	15
213	Pathogenicity and functional impact of non-frameshifting insertion/deletion variation in the human genome. *PLoS Computational Biology*, **2019**, 15, e1007112	5	15
212	polyadenylation signal variants cause syndromic microphthalmia. *Journal of Medical Genetics*, **2019**, 56, 444-452	5.8	15
211	Evaluation of copy number variation and gene expression in neurofibromatosis type-1-associated malignant peripheral nerve sheath tumours. *Human Genomics*, **2015**, 9, 3	6.8	15
210	Genomic variants in the FTO gene are associated with sporadic amyotrophic lateral sclerosis in Greek patients. *Human Genomics*, **2017**, 11, 30	6.8	15
209	A Role for Non-B DNA Forming Sequences in Mediating Microlesions Causing Human Inherited Disease. *Human Mutation*, **2016**, 37, 65-73	4.7	15

208	Application of Economic Evaluation to Assess Feasibility for Reimbursement of Genomic Testing as Part of Personalized Medicine Interventions. *Frontiers in Pharmacology*, **2019**, 10, 830	5.6	15
207	Improving the in silico assessment of pathogenicity for compensated variants. *European Journal of Human Genetics*, **2016**, 25, 2-7	5.3	15
206	Analysis of crossover breakpoints yields new insights into the nature of the gene conversion events associated with large NF1 deletions mediated by nonallelic homologous recombination. *Human Mutation*, **2014**, 35, 215-26	4.7	15
205	Assessing the pathological relevance of SPINK1 promoter variants. *European Journal of Human Genetics*, **2011**, 19, 1066-73	5.3	15
204	Legal and ethical consequences of international biobanking from a national perspective: the German BMB-EUCoop project. *European Journal of Human Genetics*, **2010**, 18, 522-5	5.3	15
203	Molecular genetic approaches to the analysis and diagnosis of human inherited disease: an overview. *Annals of Medicine*, **1992**, 24, 29-42	1.5	15
202	The distribution of the dinucleotide CpG and cytosine methylation in the vitellogenin gene family. *Journal of Molecular Evolution*, **1987**, 25, 107-15	3.1	15
201	No significant enrichment of rare functionally defective CPA1 variants in a large Chinese idiopathic chronic pancreatitis cohort. *Human Mutation*, **2017**, 38, 959-963	4.7	14
200	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	2.5	14
199	The somatic autosomal mutation matrix in cancer genomes. *Human Genetics*, **2015**, 134, 851-64	6.3	14
198	Technological advances in DNA sequence enrichment and sequencing for germline genetic diagnosis. *Expert Review of Molecular Diagnostics*, **2012**, 12, 159-73	3.8	14
197	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	4.4	14
196	Polymorphic micro-inversions contribute to the genomic variability of humans and chimpanzees. *Human Genetics*, **2006**, 119, 103-12	6.3	14
195	Disentangling the perturbational effects of amino acid substitutions in the DNA-binding domain of p53. *Human Genetics*, **1999**, 104, 15-22	6.3	14
194	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, 351-70	6.3	14
193	Protein C London 1: recurrent mutation at Arg 169 (CGG----TGG) in the protein C gene causing thrombosis. *Nucleic Acids Research*, **1989**, 17, 10513	20.1	14
192	MutationTaster2021. *Nucleic Acids Research*, **2021**, 49, W446-W451	20.1	14
191	First estimate of the scale of canonical 5' splice site GT>GC variants capable of generating wild-type transcripts. *Human Mutation*, **2019**, 40, 1856-1873	4.7	13

190	The Rise and Rise of Exome Sequencing. *Public Health Genomics*, **2016**, 19, 315-324	1.9	13
189	Fine mapping of meiotic NAHR-associated crossovers causing large NF1 deletions. *Human Molecular Genetics*, **2016**, 25, 484-96	5.6	13
188	Structure-based kernels for the prediction of catalytic residues and their involvement in human inherited disease. *Bioinformatics*, **2010**, 26, 1975-82	7.2	13
187	Enigmatic in vivo iduronate-2-sulfatase (IDS) mutant transcript correction to wild-type in Hunter syndrome. *Human Mutation*, **2010**, 31, E1261-85	4.7	13
186	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	4	13
185	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	6.3	13
184	A list of cloned human DNA sequences. *Human Genetics*, **1983**, 65, 19-26	6.3	13
183	A list of cloned human DNA sequences--supplement. *Human Genetics*, **1984**, 67, 111-4	6.3	13
182	Identification of a functional enhancer variant within the chronic pancreatitis-associated SPINK1 c.101A>G (p.Asn34Ser)-containing haplotype. *Human Mutation*, **2017**, 38, 1014-1024	4.7	12
181	Regulatory Single-Nucleotide Variant Predictor Increases Predictive Performance of Functional Regulatory Variants. *Human Mutation*, **2016**, 37, 1137-1143	4.7	12
180	Phenotypic and genotypic overlap between mosaic NF2 and schwannomatosis in patients with multiple non-intradermal schwannomas. *Human Genetics*, **2018**, 137, 543-552	6.3	12
179	A new and more accurate estimate of the rate of concurrent tandem-base substitution mutations in the human germline: ~0.4% of the single-nucleotide substitution mutation rate. *Human Mutation*, **2014**, 35, 392-4	4.7	12
178	An emerging role for microRNAs in NF1 tumorigenesis. *Human Genomics*, **2012**, 6, 23	6.8	12
177	Gene conversion in human genetic disease. *Genes*, **2010**, 1, 550-63	4.2	12
176	A gene conversion hotspot in the human growth hormone (GH1) gene promoter. *Human Mutation*, **2009**, 30, 239-47	4.7	12
175	Triangulation of the human, chimpanzee, and Neanderthal genome sequences identifies potentially compensated mutations. *Human Mutation*, **2010**, 31, 1286-93	4.7	12
174	Mechanism of Alu integration into the human genome. *Genomic Medicine*, **2007**, 1, 9-17		12
173	Single-strand conformation polymorphism (SSCP) analysis of exon 11 of the CFTR gene reliably detects more than one third of non-delta F508 mutations in German cystic fibrosis patients. *Human Genetics*, **1992**, 88, 283-7	6.3	12

172	The pattern of DNA methylation in the delta-crystallin genes in transdifferentiating neural retina cultures. *Differentiation*, **1983**, 24, 33-8	3.5	12
171	AMELIE accelerates Mendelian patient diagnosis directly from the primary literature		12
170	AVADA: toward automated pathogenic variant evidence retrieval directly from the full-text literature. *Genetics in Medicine*, **2020**, 22, 362-370	8.1	12
169	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	12
168	Severe infantile isolated exocrine pancreatic insufficiency caused by the complete functional loss of the SPINK1 gene. *Human Mutation*, **2017**, 38, 1660-1665	4.7	11
167	AMELIE speeds Mendelian diagnosis by matching patient phenotype and genotype to primary literature. *Science Translational Medicine*, **2020**, 12,	17.5	11
166	Local DNA dynamics shape mutational patterns of mononucleotide repeats in human genomes. *Nucleic Acids Research*, **2015**, 43, 5065-80	20.1	11
165	How to distinguish genetically between an alleged father and his monozygotic twin: a thought experiment. *Forensic Science International: Genetics*, **2012**, 6, e129-30	4.3	11
164	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	3.7	11
163	Delineating the Hemostaseome as an aid to individualize the analysis of the hereditary basis of thrombotic and bleeding disorders. *Human Genetics*, **2011**, 130, 149-66	6.3	11
162	Comparative analysis of germline and somatic microlesion mutational spectra in 17 human tumor suppressor genes. *Human Mutation*, **2011**, 32, 620-32	4.7	11
161	Prenatal exclusion of severe factor VII deficiency. *Journal of Pediatric Hematology/Oncology*, **2003**, 25, 418-20	1.2	11
160	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	6.3	11
159	The molecular genetics of familial venous thrombosis. *Best Practice and Research: Clinical Haematology*, **1994**, 7, 637-74		11
158	The molecular genetics of familial venous thrombosis. *Blood Reviews*, **1991**, 5, 55-70	11.1	11
157	The Loss and Gain of Functional Amino Acid Residues Is a Common Mechanism Causing Human Inherited Disease. *PLoS Computational Biology*, **2016**, 12, e1005091	5	11
156	Developmental Gene Expression Differences between Humans and Mammalian Models. *Cell Reports*, **2020**, 33, 108308	10.6	11
155	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	6.3	11

154	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	2	11
153	Biological and functional relevance of CASP predictions. *Proteins: Structure, Function and Bioinformatics*, **2018**, 86 Suppl 1, 374-386	4.2	10
152	ExonImpact: Prioritizing Pathogenic Alternative Splicing Events. *Human Mutation*, **2017**, 38, 16-24	4.7	10
151	Research and clinical applications of cancer genome sequencing. *Current Opinion in Obstetrics and Gynecology*, **2013**, 25, 3-10	2.4	10
150	Delineation of the clinical phenotype associated with non-mosaic type-2 NF1 deletions: two case reports. *Journal of Medical Case Reports*, **2011**, 5, 577	1.2	10
149	regSNPs: a strategy for prioritizing regulatory single nucleotide substitutions. *Bioinformatics*, **2012**, 28, 1879-86	7.2	10
148	Molecular analysis of the 5'-flanking region of the neurofibromatosis type 1 (NF1) gene: identification of five sequence variants. *Clinical Genetics*, **2000**, 57, 221-4	4	10
147	Molecular genetic analysis of severe protein C deficiency. *Human Genetics*, **2000**, 106, 646-653	6.3	10
146	Screening for mutations in the antithrombin III gene causing recurrent venous thrombosis by single-strand conformation polymorphism analysis. *Human Mutation*, **1993**, 2, 324-6	4.7	10
145	Nucleotide Weight Matrices Reveal Ubiquitous Mutational Footprints of AID/APOBEC Deaminases in Human Cancer Genomes. *Cancers*, **2019**, 11,	6.6	9
144	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	11.5	9
143	RegulationSpotter: annotation and interpretation of extratranscriptic DNA variants. *Nucleic Acids Research*, **2019**, 47, W106-W113	20.1	9
142	Chromosomal speciation of humans and chimpanzees revisited: studies of DNA divergence within inverted regions. *Cytogenetic and Genome Research*, **2007**, 116, 53-60	1.9	9
141	NF1 Microdeletions and Their Underlying Mutational Mechanisms **2012**, 187-209		9
140	iRegNet3D: three-dimensional integrated regulatory network for the genomic analysis of coding and non-coding disease mutations. *Genome Biology*, **2017**, 18, 10	18.3	8
139	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	20.1	8
138	Pronounced maternal parent-of-origin bias for type-1 NF1 microdeletions. *Human Genetics*, **2018**, 137, 365-373	6.3	8
137	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	6.8	8

136	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, 206-10	2.9	8
135	Complex Multiple-Nucleotide Substitution Mutations Causing Human Inherited Disease Reveal Novel Insights into the Action of Translesion Synthesis DNA Polymerases. *Human Mutation*, **2015**, 36, 1034-8	4.7	8
134	Genetics in genomic era. *Genetics Research International*, **2015**, 2015, 364960	0	8
133	Concurrent nucleotide substitution mutations in the human genome are characterized by a significantly decreased transition/transversion ratio. *Human Mutation*, **2015**, 36, 333-41	4.7	8
132	A probabilistic model to predict clinical phenotypic traits from genome sequencing. *PLoS Computational Biology*, **2014**, 10, e1003825	5	8
131	The mutational demography of protein C deficiency. *Human Genetics*, **1995**, 96, 142-6	6.3	8
130	Core database. *Nature*, **1995**, 374, 402	50.4	8
129	Regional localization and characterization of a DNA segment on the long arm of chromosome 21. *Human Genetics*, **1987**, 75, 129-35	6.3	8
128	Consideration of the haplotype diversity at nonallelic homologous recombination hotspots improves the precision of rearrangement breakpoint identification. *Human Mutation*, **2017**, 38, 1711-1722	4.7	8
127	In vitro and in silico evidence against a significant effect of the c.194G>A variant on pre-mRNA splicing. *Gut*, **2017**, 66, 2195-2196	19.2	7
126	copy number variants and promoter polymorphisms in pancreatitis: common pathogenetic mechanism, different genetic effects. *Gut*, **2018**, 67, 592-593	19.2	7
125	IMHOTEP-a composite score integrating popular tools for predicting the functional consequences of non-synonymous sequence variants. *Nucleic Acids Research*, **2017**, 45, e13	20.1	7
124	Extreme clustering of type-1 NF1 deletion breakpoints co-locating with G-quadruplex forming sequences. *Human Genetics*, **2018**, 137, 511-520	6.3	7
123	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, 384-93	6.8	7
122	Extended runs of homozygosity at 17q11.2: an association with type-2 NF1 deletions?. *Human Mutation*, **2010**, 31, 325-34	4.7	7
121	Variation of site-specific methylation patterns in the factor VIII (F8C) gene in human sperm DNA. *Human Genetics*, **1998**, 103, 228-33	6.3	7
120	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**, 131, 227-31		7
119	The evolution of the vertebrate beta-globin gene promoter. *Evolution; International Journal of Organic Evolution*, **2002**, 56, 224-32	3.8	7

118	Protein C deficiency and thromboembolism: recurrent mutation at Arg 306 in the protein C gene. *Human Genetics*, **1992**, 88, 586-8	6.3	7
117	Molecular genetic analysis of a novel form of haemophilia A characterized by the variable expression of factor VIII. *Thrombosis Research*, **1990**, 59, 871-7	8.2	7
116	Human gene cloning: the storm before the lull?. *Nature*, **1986**, 322, 119	50.4	7
115	Non-coding RNA ANRIL and the number of plexiform neurofibromas in patients with NF1 microdeletions. *BMC Medical Genetics*, **2012**, 13, 98	2.1	6
114	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, 201-3	4	6
113	Prothrombin cleavage by human vascular smooth muscle cells: a potential alternative pathway to the coagulation cascade. *Journal of Cellular Biochemistry*, **1995**, 59, 514-28	4.7	6
112	A comprehensive list of cloned human DNA sequences. *Nucleic Acids Research*, **1990**, 18 Suppl, 2413-547	20.1	6
111	Identification and functional characterization of a novel heterozygous missense variant in the LPL associated with recurrent hypertriglyceridemia-induced acute pancreatitis in pregnancy. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1048	2.3	5
110	Mining clinical attributes of genomic variants through assisted literature curation in Egas. *Database: the Journal of Biological Databases and Curation*, **2016**, 2016,	5	5
109	Digging deeper into the intronic sequences of the SPINK1 gene. *Gut*, **2016**, 65, 1055-6	19.2	5
108	CDG: An Online Server for Detecting Biologically Closest Disease-Causing Genes and its Application to Primary Immunodeficiency. *Frontiers in Immunology*, **2018**, 9, 1340	8.4	5
107	Identification of large NF1 duplications reciprocal to NAHR-mediated type-1 NF1 deletions. *Human Mutation*, **2014**, 35, 1469-75	4.7	5
106	In silico prioritization and further functional characterization of SPINK1 intronic variants. *Human Genomics*, **2017**, 11, 7	6.8	5
105	Disclosing the Hidden Structure and Underlying Mutational Mechanism of a Novel Type of Duplication CNV Responsible for Hereditary Multiple Osteochondromas. *Human Mutation*, **2015**, 36, 758-63	4.7	5
104	Human Gene Mutation: Mechanisms and Consequences **2010**, 319-363		5
103	Homology modelling of the catalytic domain of early mammalian protein C: evolution of structural features. *Human Genetics*, **1997**, 101, 37-42	6.3	5
102	Searching for potential microRNA-binding site mutations amongst known disease-associated 3' UTR variants. *Genomic Medicine*, **2007**, 1, 29-33		5
101	A single base-pair deletion in the protein C gene causing recurrent thromboembolism. *Thrombosis Research*, **1991**, 61, 335-40	8.2	5

100	The Experimentally Obtained Functional Impact Assessments of 5' Splice Site GT'GC Variants Differ Markedly from Those Predicted. *Current Genomics*, **2020**, 21, 56-66	2.6	5
99	Neuroprotectants attenuate hypobaric hypoxia-induced brain injuries in cynomolgus monkeys. *Zoological Research*, **2020**, 41, 3-19	3.4	5
98	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	7	5
97	Role of the Common Haplotype in Alcoholic and Non-Alcoholic Chronic Pancreatitis: Meta- and Re-Analyses. *Genes*, **2020**, 11,	4.2	5
96	Toward a clinical diagnostic pipeline for SPINK1 intronic variants. *Human Genomics*, **2019**, 13, 8	6.8	5
95	Intra-individual plasticity of the TAZ gene leading to different heritable mutations in siblings with Barth syndrome. *European Journal of Human Genetics*, **2015**, 23, 1708-12	5.3	4
94	5' splice site GC>GT and GT>GC variants differ markedly in terms of their functionality and pathogenicity. *Human Mutation*, **2020**, 41, 1358-1364	4.7	4
93	Quantitative mapping of genetic similarity in human heritable diseases by shared mutations. *Human Mutation*, **2018**, 39, 292-301	4.7	4
92	Screening in silico predicted remotely acting NF1 gene regulatory elements for mutations in patients with neurofibromatosis type 1. *Human Genomics*, **2013**, 7, 18	6.8	4
91	DNA structure matters. *Genome Medicine*, **2013**, 5, 51	14.4	4
90	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,	4.2	4
89	'Sifting the significance from the data' - the impact of high-throughput genomic technologies on human genetics and health care. *Human Genomics*, **2012**, 6, 11	6.8	4
88	The Germline Mutational Spectrum in Neurofibromatosis Type 1 and Genotype□Phenotype Correlations **2012**, 115-134		4
87	Lionizing lyonization 50□years on. *Human Genetics*, **2011**, 130, 167-8	6.3	4
86	Do inherited disease genes have distinguishing functional characteristics?. *Genetic Testing and Molecular Biomarkers*, **2010**, 14, 289-91	1.6	4
85	Two sisters with Rett syndrome and non-identical paternally-derived microdeletions in the MECP2 gene. *Genomic Medicine*, **2008**, 2, 77-81		4
84	A rare complex DNA rearrangement in the murine Steel gene results in exon duplication and a lethal phenotype. *Blood*, **2003**, 102, 3548-55	2.2	4
83	The Frequency of Inherited Disorders Database. *Human Genetics*, **2001**, 108, 72-4	6.3	4

82	A novel missense mutation in the antithrombin III gene (Ser349----Pro) causing recurrent venous thrombosis. *Human Genetics*, **1992**, 88, 707-8	6.3	4
81	S-CAP extends clinical-grade pathogenicity prediction to genetic variants that affect RNA splicing		4
80	Verifying nomenclature of DNA variants in submitted manuscripts: Guidance for journals. *Human Mutation*, **2021**, 42, 3-7	4.7	4
79	A platform for curated products from novel open reading frames prompts reinterpretation of disease variants. *Genome Research*, **2021**,	9.7	4
78	Discovery and Functional Annotation of PRSS1 Promoter Variants in Chronic Pancreatitis. *Human Mutation*, **2016**, 37, 1149-1152	4.7	3
77	Analysis of Features from Protein-protein Hetero-complex Structures to Predict Protein Interaction Interfaces Using Machine Learning. *Procedia Technology*, **2013**, 10, 62-66		3
76	Cross-comparison of the genome sequences from human, chimpanzee, Neanderthal and a Denisovan hominin identifies novel potentially compensated mutations. *Human Genomics*, **2011**, 5, 453-84	6.8	3
75	Local DNA sequence determinants of FUT2 copy number variation. *Transfusion*, **2011**, 51, 1359-61	2.9	3
74	Prospects for the automated extraction of mutation data from the scientific literature. *Human Genomics*, **2010**, 5, 1-4	6.8	3
73	Is the NIH policy for sharing GWAS data running the risk of being counterproductive?. *Investigative Genetics*, **2010**, 1, 3		3
72	Comparative analysis of copy number variation in primate genomes. *Cytogenetic and Genome Research*, **2008**, 123, 288-96	1.9	3
71	Compound heterozygosity for two novel mutations (1203insG/Y1456X) in the von Willebrand factor gene causing type 3 von Willebrand disease. *Haemophilia*, **2007**, 13, 645-8	3.3	3
70	Detection of NF1 mutations utilizing the protein truncation test (PTT). *Methods in Molecular Biology* , **2003**, 217, 315-27	1.4	3
69	MspI RFLP in the human heparin cofactor II (HCF2) gene. *Nucleic Acids Research*, **1990**, 18, 1664	20.1	3
68	Molecular genetic approaches to the analysis of human ophthalmic disease. *Eye*, **1987**, 1 (Pt 6), 699-721	4.4	3
67	Mutations Causing Complex Disease May under Certain Circumstances Be Protective in an Epidemiological Sense. *PLoS ONE*, **2015**, 10, e0132150	3.7	3
66	Atypical Microdeletions: Challenges and Opportunities for Genotype/Phenotype Correlations in Patients with Large Deletions. *Genes*, **2021**, 12,	4.2	3
65	The Somatic Mutational Spectrum of the NF1 Gene **2012**, 211-233		3

64	Pathogenic and likely pathogenic variants in at least five genes account for approximately 3% of mild isolated nonsyndromic thrombocytopenia. *Transfusion*, **2020**, 60, 2419-2431	2.9	3
63	NGS mismapping confounds the clinical interpretation of the p.Ala16Val (c.47C>T) variant in chronic pancreatitis. *Gut*, **2021**,	19.2	3
62	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,	11.5	3
61	Classification of NF1 microdeletions and its importance for establishing genotype/phenotype correlations in patients with NF1 microdeletions. *Human Genetics*, **2021**, 140, 1635-1649	6.3	3
60	Gene Conversion in Evolution and Disease		3
59	Compound Heterozygosity for Novel Truncating Variants in the Gene as the Cause of Polyhydramnios in Two Successive Fetuses. *Frontiers in Genetics*, **2019**, 10, 835	4.5	2
58	and 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	4.5	2
57	Gene-environment interaction between APOA5 c.553G>T and pregnancy in hypertriglyceridemia-induced acute pancreatitis. *Journal of Clinical Lipidology*, **2020**, 14, 498-506	4.9	2
56	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	4.3	2
55	Chromosomal distribution of disease genes in the human genome. *Genetic Testing and Molecular Biomarkers*, **2010**, 14, 441-6	1.6	2
54	Structure-based kernels for the prediction of catalytic residues and their involvement in human inherited disease. *BMC Bioinformatics*, **2010**, 11, O4	3.6	2
53	Copy number variation and disease. Preface. *Cytogenetic and Genome Research*, **2008**, 123, 5-6	1.9	2
52	Mutations in Human Genetic Disease **2006**,		2
51	A comprehensive list of cloned human DNA sequences--1990 update. *Nucleic Acids Research*, **1991**, 19 Suppl, 2111-26	20.1	2
50	Prenatal exclusion of haemophilia A and carrier testing by direct detection of a disease lesion. *Prenatal Diagnosis*, **1992**, 12, 861-6	3.2	2
49	Analysis of missense variants in the human genome reveals widespread gene-specific clustering and improves prediction of pathogenicity.. *American Journal of Human Genetics*, **2022**,	11	2
48	PIVOTAL: Prioritizing variants of uncertain significance with spatial genomic patterns in the 3D proteome		2
47	Developmental gene expression differences between humans and mammalian models		2

46	Common polymorphic OTC variants can act as genetic modifiers of enzymatic activity. *Human Mutation*, **2021**, 42, 978-989	4.7	2
45	Scale and Scope of Gene-Alcohol Interactions in Chronic Pancreatitis: A Systematic Review. *Genes*, **2021**, 12,	4.2	2
44	The sequencing and interpretation of the genome obtained from a Serbian individual. *PLoS ONE*, **2018**, 13, e0208901	3.7	2
43	Human Gene Mutation in Inherited Disease **2013**, 1-48		1
42	Sequential data selection for predicting the pathogenic effects of sequence variation **2015**,		1
41	Local sequence determinants of two in-frame triplet deletion/duplication hotspots in the RHD/RHCE genes. *Human Genomics*, **2012**, 6, 8	6.8	1
40	The Sequencing of the Rhesus Macaque Genome and its Comparison with the Genome Sequences of Human and Chimpanzee **2008**,		1
39	Neurofibromatosis Type 1 **2004**, 285-310		1
38	THE EVOLUTION OF THE VERTEBRATE □GLOBIN GENE PROMOTER. *Evolution; International Journal of Organic Evolution*, **2002**, 56, 224	3.8	1
37	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	6.3	1
36	Carrier detection in haemophilia A by direct analysis of factor VIII gene lesions. *Human Genetics*, **1991**, 87, 99-100	6.3	1
35	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	11	1
34	RegSNPs-Intron: A computational framework for prioritizing Intronic Single Nucleotide Variants in Human Genetic Disease		1
33	5□splice site GC>GT variants differ from GT>GC variants in terms of their functionality and pathogenicity		1
32	AVADA Enables Automated Genetic Variant Curation Directly from the Full Text Literature		1
31	Genetic analysis of the STIM1 gene in chronic pancreatitis		1
30	AMELIE 2 speeds up Mendelian diagnosis by matching patient phenotype & genotype to primary literature		1
29	Trypsinogen Genes: Insights into Molecular Evolution from the Study of Pathogenic Mutations		1

28	Digenic Inheritance and Gene-Environment Interaction in a Patient With Hypertriglyceridemia and Acute Pancreatitis. *Frontiers in Genetics*, **2021**, 12, 640859	4.5	1
27	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	4.5	1
26	Compensatory epistasis explored by molecular dynamics simulations. *Human Genetics*, **2021**, 140, 1329-1342	6.3	1
25	Most unambiguous loss-of-function mutations are unlikely to predispose to chronic pancreatitis. *Gut*, **2020**, 69, 785-786	19.2	1
24	The Genomic Medicine Alliance: A Global Effort to Facilitate the Introduction of Genomics into Healthcare in Developing Nations **2018**, 173-188		1
23	Splicing Outcomes of 5' 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	4.5	1
22	De Novo Mutations in Human Inherited Disease1-7		1
21	Trypsinogen (PRSS1 and PRSS2) gene dosage correlates with pancreatitis risk across genetic and transgenic studies: a systematic review and re-analysis.. *Human Genetics*, **2022**, 1	6.3	0
20	Non-B DNA Structure and Mutations Causing Human Genetic Disease1-15		0
19	Human Genomic Variants and Inherited Disease: Molecular Mechanisms and Clinical Consequences **2019**, 125-200		0
18	The reversion variant (p.Arg90Leu) at the evolutionarily adaptive p.Arg90 site in CELA3B predisposes to chronic pancreatitis. *Human Mutation*, **2021**, 42, 385-391	4.7	0
17	Prioritization of schizophrenia risk genes from GWAS results by integrating multi-omics data. *Translational Psychiatry*, **2021**, 11, 175	8.6	0
16	No Convincing Evidence to Support a Bimodal Age of Onset in Idiopathic Chronic Pancreatitis. *Clinical Gastroenterology and Hepatology*, **2021**,	6.9	0
15	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*, **2021**, 141, 177	6.3	0
14	The Evolution of High-Throughput Sequencing Technologies: From Sanger to Single-Molecule Sequencing **2013**, 1-30		
13	Next-generation sequencing in cancer research & diagnostics **2013**, 20-40		
12	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	4.7	
11	Remotely acting SMCHD1 gene regulatory elements: in silico prediction and identification of potential regulatory variants in patients with FSHD. *Human Genomics*, **2015**, 9, 25	6.8	

10 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

9 [14]Analysis of promoter mutations causing human genetic disease. *Methods in Molecular Genetics*, **1996**, 8, 261-277

8 The Molecular Genetics of Platelet Membrane Proteins and their Inherited Disorders. *Platelets*, **1991**, 2, 59-67 3.6

7 Structure and function in the human genome **2020**, 1-41

6 Mapping the human genome **2020**, 43-68

5 Application of PCR to the Detection and Analysis of Point Mutations in the Human Factor VIII Gene **1991**, 23-31

4 Sequencing the Human Genome: Novel Insights into Its Structure and Function1-9

3 Pathogenetics of Chronic Pancreatitis **2017**, 63-77

2 Somatic Copy Number Alterations: Gene and Protein Expression Correlates in NF1-Associated Malignant Peripheral Nerve Sheath Tumors **2012**, 405-428

1 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 1.6