# David N Cooper

List of Publications by Year
in descending order



Version: 2024-02-01

| | | | |
|---|---|---|---|
| 469 papers | 83,715 citations | 2318 98 h-index | 515 267 g-index |
| 523 all docs | 523 docs citations | 523 times ranked | 95086 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. | 6.1 | 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. | 1.8 | 8 |
| 3 | Distinct sequence features underlie microdeletions and gross deletions in the human genome. Human Mutation, 2022, 43, 328-346. | 1.1 | 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. | 2.6 | 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. | 2.3 | 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. | 1.8 | 29 |
| 7 | Functionally deficient TRPV6 variants contribute to hereditary and familial chronic pancreatitis. Human Mutation, 2022, 43, 228-239. | 1.1 | 7 |
| 8 | Verifying nomenclature of DNA variants in submitted manuscripts: Guidance for journals. Human Mutation, 2021, 42, 3-7. | 1.1 | 10 |
| 9 | A platform for curated products from novel open reading frames prompts reinterpretation of disease variants. Genome Research, 2021, 31, 327-336. | 2.4 | 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. | 1.1 | 6 |
| 11 | Scale and Scope of Gene-Alcohol Interactions in Chronic Pancreatitis: A Systematic Review. Genes, 2021, 12, 471. | 1.0 | 9 |
| 12 | Prioritization of schizophrenia risk genes from GWAS results by integrating multi-omics data. Translational Psychiatry, 2021, 11, 175. | 2.4 | 10 |
| 13 | Digenic Inheritance and Gene-Environment Interaction in a Patient With Hypertriglyceridemia and Acute Pancreatitis. Frontiers in Genetics, 2021, 12, 640859. | 1.1 | 7 |
| 14 | MutationTaster2021. Nucleic Acids Research, 2021, 49, W446-W451. | 6.5 | 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. | 1.1 | 4 |
| 16 | Compensatory epistasis explored by molecular dynamics simulations. Human Genetics, 2021, 140, 1329-1342. | 1.8 | 6 |
| 17 | Common polymorphic OTC variants can act as genetic modifiers of enzymatic activity. Human Mutation, 2021, 42, 978-989. | 1.1 | 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, . | 3.3 | 42 |

| # | Article | IF | Citations |
|---|---|---|---|
| 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. | 1.1 | 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. | 1.8 | 12 |
| 21 | No Convincing Evidence to Support a Bimodal Age of Onset in Idiopathic Chronic Pancreatitis. Clinical Gastroenterology and Hepatology, 2021, , . | 2.4 | 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. | 6.5 | 29 |
| 23 | Chronic Pancreatitis: The True Pathogenic Culprit within the SPINK1 N34S-Containing Haplotype Is No Longer at Large. Genes, 2021, 12, 1683. | 1.0 | 5 |
| 24 | Atypical NF1 Microdeletions: Challenges and Opportunities for Genotype/Phenotype Correlations in Patients with Large NF1 Deletions. Genes, 2021, 12, 1639. | 1.0 | 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. | 2.6 | 21 |
| 26 | Most unambiguous loss-of-function CPA1 mutations are unlikely to predispose to chronic pancreatitis. Gut, 2020, 69, 785-786. | 6.1 | 6 |
| 27 | AVADA: toward automated pathogenic variant evidence retrieval directly from the full-text literature. Genetics in Medicine, 2020, 22, 362-370. | 1.1 | 24 |
| 28 | Role of the Common PRSS1-PRSS2 Haplotype in Alcoholic and Non-Alcoholic Chronic Pancreatitis: Meta- and Re-Analyses. Genes, 2020, 11, 1349. | 1.0 | 14 |
| 29 | Inferring the molecular and phenotypic impact of amino acid variants with MutPred2. Nature Communications, 2020, 11, 5918. | 5.8 | 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. | 0.8 | 6 |
| 31 | Developmental Gene Expression Differences between Humans and Mammalian Models. Cell Reports, 2020, 33, 108308. | 2.9 | 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. | 1.1 | 4 |
| 33 | AMELIE speeds Mendelian diagnosis by matching patient phenotype and genotype to primary literature. Science Translational Medicine, 2020, 12, . | 5.8 | 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. | 1.1 | 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. | 3.3 | 18 |
| 36 | The Human Gene Mutation Database (HGMDÂ®): optimizing its use in a clinical diagnostic or research setting. Human Genetics, 2020, 139, 1197-1207. | 1.8 | 353 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Geneâ€“environment interaction between APOA5Âc.553G>T and pregnancy in hypertriglyceridemia-induced acute pancreatitis. Journal of Clinical Lipidology, 2020, 14, 498-506. | 0.6 | 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. | 0.6 | 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. | 0.7 | 16 |
| 40 | Neuroprotectants attenuate hypobaric hypoxia-induced brain injuries in cynomolgus monkeys. Zoological Research, 2020, 41, 3-19. | 0.9 | 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. | 1.6 | 26 |
| 44 | RegulationSpotter: annotation and interpretation of extratranscriptic DNA variants. Nucleic Acids Research, 2019, 47, W106-W113. | 6.5 | 17 |
| 45 | Uganda Genome Resource Enables Insights into Population History and Genomic Discovery in Africa. Cell, 2019, 179, 984-1002.e36. | 13.5 | 152 |
| 46 | Extensive disruption of protein interactions by genetic variants across the allele frequency spectrum in human populations. Nature Communications, 2019, 10, 4141. | 5.8 | 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. | 1.1 | 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. | 1.1 | 25 |
| 49 | Gene expression across mammalian organ development. Nature, 2019, 571, 505-509. | 13.7 | 490 |
| 50 | Pathogenicity and functional impact of non-frameshifting insertion/deletion variation in the human genome. PLoS Computational Biology, 2019, 15, e1007112. | 1.5 | 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. | 6.5 | 15 |
| 52 | NAA10 polyadenylation signal variants cause syndromic microphthalmia. Journal of Medical Genetics, 2019, 56, 444-452. | 1.5 | 28 |
| 53 | Nucleotide Weight Matrices Reveal Ubiquitous Mutational Footprints of AID/APOBEC Deaminases in Human Cancer Genomes. Cancers, 2019, 11, 211. | 1.7 | 15 |
| 54 | S-CAP extends pathogenicity prediction to genetic variants that affect RNA splicing. Nature Genetics, 2019, 51, 755-763. | 9.4 | 56 |

| # | Article | IF | Citations |
|---|---|---|---|
| 55 | Toward a clinical diagnostic pipeline for SPINK1 intronic variants. Human Genomics, 2019, 13, 8. | 1.4 | 8 |
| 56 | RegSNPs-intron: a computational framework for predicting pathogenic impact of intronic single nucleotide variants. Genome Biology, 2019, 20, 254. | 3.8 | 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. | 3.3 | 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. | 1.8 | 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. | 0.7 | 23 |
| 61 | Pronounced maternal parent-of-origin bias for type-1 NF1 microdeletions. Human Genetics, 2018, 137, 365-373. | 1.8 | 12 |
| 62 | PRSS1 copy number variants and promoter polymorphisms in pancreatitis: common pathogenetic mechanism, different genetic effects. Gut, 2018, 67, 592-593. | 6.1 | 12 |
| 63 | Quantitative mapping of genetic similarity in human heritable diseases by shared mutations. Human Mutation, 2018, 39, 292-301. | 1.1 | 8 |
| 64 | FATHMM-XF: accurate prediction of pathogenic point mutations via extended features. Bioinformatics, 2018, 34, 511-513. | 1.8 | 296 |
| 65 | Biological and functional relevance of CASP predictions. Proteins: Structure, Function and Bioinformatics, 2018, 86, 374-386. | 1.5 | 12 |
| 66 | DNA polymerase Î· mutational signatures are found in a variety of different types of cancer. Cell Cycle, 2018, 17, 348-355. | 1.3 | 32 |
| 67 | SPINK1 , PRSS1 , CTRC , and CFTR Genotypes Influence Disease Onset and Clinical Outcomes in Chronic Pancreatitis. Clinical and Translational Gastroenterology, 2018, 9, e204. | 1.3 | 76 |
| 68 | The sequencing and interpretation of the genome obtained from a Serbian individual. PLoS ONE, 2018, 13, e0208901. | 1.1 | 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. | 2.2 | 6 |
| 72 | Phenotypic and genotypic overlap between mosaic NF2 and schwannomatosis in patients with multiple non-intradermal schwannomas. Human Genetics, 2018, 137, 543-552. | 1.8 | 25 |

| # | Article | IF | Citations |
|---|---|---|---|
| 73 | Extreme clustering of type-1 NF1 deletion breakpoints co-locating with G-quadruplex forming sequences. Human Genetics, 2018, 137, 511-520. | 1.8 | 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. | 1.4 | 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. | 6.5 | 10 |
| 76 | iRegNet3D: three-dimensional integrated regulatory network for the genomic analysis of coding and non-coding disease mutations. Genome Biology, 2017, 18, 10. | 3.8 | 9 |
| 77 | Emerging genotypeâ€"phenotype relationships in patients with large NF1 deletions. Human Genetics, 2017, 136, 349-376. | 1.8 | 163 |
| 78 | regSNPs-splicing: a tool for prioritizing synonymous single-nucleotide substitution. Human Genetics, 2017, 136, 1279-1289. | 1.8 | 27 |
| 79 | Mutational signatures and mutable motifs in cancer genomes. Briefings in Bioinformatics, 2017, 19, 1085-1101. | 3.2 | 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. | 1.1 | 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. | 1.1 | 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. | 1.8 | 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. | 6.1 | 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. | 1.8 | 106 |
| 85 | Severe infantile isolated exocrine pancreatic insufficiency caused by the complete functional loss of theSPINK1gene. Human Mutation, 2017, 38, 1660-1665. | 1.1 | 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. | 1.1 | 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. | 1.0 | 29 |
| 88 | Investigating DNA-, RNA-, and protein-based features as a means to discriminate pathogenic synonymous variants. Human Mutation, 2017, 38, 1336-1347. | 1.1 | 37 |
| 89 | In silico prioritization and further functional characterization of SPINK1 intronic variants. Human Genomics, 2017, 11, 7. | 1.4 | 10 |
| 90 | The NF1 somatic mutational landscape in sporadic human cancers. Human Genomics, 2017, 11, 13. | 1.4 | 203 |

| # | Article | IF | Citations |
|---|---|---|---|
| 91 | ExonImpact: Prioritizing Pathogenic Alternative Splicing Events. Human Mutation, 2017, 38, 16-24. | 1.1 | 12 |
| 92 | Improving the in silico assessment of pathogenicity for compensated variants. European Journal of Human Genetics, 2017, 25, 2-7. | 1.4 | 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. | 1.8 | 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. | 1.0 | 10 |
| 95 | Genomic variants in the FTO gene are associated with sporadic amyotrophic lateral sclerosis in Greek patients. Human Genomics, 2017, 11, 30. | 1.4 | 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. | 1.2 | 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. | 1.1 | 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. | 1.1 | 94 |
| 100 | Test Pricing and Reimbursement in Genomic Medicine: Towards a General Strategy. Public Health Genomics, 2016, 19, 352-363. | 0.6 | 37 |
| 101 | No Association Between CELâ€“HYB Hybrid Allele and Chronic Pancreatitis in Asian Populations. Gastroenterology, 2016, 150, 1558-1560.e5. | 0.6 | 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. | 6.5 | 117 |
| 103 | Analysis of protein-coding genetic variation in 60,706 humans. Nature, 2016, 536, 285-291. | 13.7 | 9,051 |
| 104 | Regulatory Single-Nucleotide Variant Predictor Increases Predictive Performance of Functional Regulatory Variants. Human Mutation, 2016, 37, 1137-1143. | 1.1 | 13 |
| 105 | Discovery and Functional Annotation ofPRSS1Promoter Variants in Chronic Pancreatitis. Human Mutation, 2016, 37, 1149-1152. | 1.1 | 5 |
| 106 | M-CAP eliminates a majority of variants of uncertain significance in clinical exomes at high sensitivity. Nature Genetics, 2016, 48, 1581-1586. | 9.4 | 654 |
| 107 | Mining clinical attributes of genomic variants through assisted literature curation in Egas. Database: the Journal of Biological Databases and Curation, 2016, 2016, baw096. | 1.4 | 6 |
| 108 | The Rise and Rise of Exome Sequencing. Public Health Genomics, 2016, 19, 315-324. | 0.6 | 15 |

| # | Article | IF | Citations |
|---|---|---|---|
| 109 | The mutation significance cutoff: gene-level thresholds for variant predictions. Nature Methods, 2016, 13, 109-110. | 9.0 | 249 |
| 110 | A Role for Non-B DNA Forming Sequences in Mediating Microlesions Causing Human Inherited Disease. Human Mutation, 2016, 37, 65-73. | 1.1 | 22 |
| 111 | Assessing the Pathogenicity of Insertion and Deletion Variants with the Variant Effect Scoring Tool (VESTâ€indel). Human Mutation, 2016, 37, 28-35. | 1.1 | 101 |
| 112 | Digging deeper into the intronic sequences of theSPINK1gene: TableÂ1. Gut, 2016, 65, 1055-1056. | 6.1 | 10 |
| 113 | Fine mapping of meiotic NAHR-associated crossovers causing largeNF1deletions. Human Molecular Genetics, 2016, 25, 484-496. | 1.4 | 15 |
| 114 | Clarifying the clinical relevance ofSPINK1intronic variants in chronic pancreatitis. Gut, 2016, 65, 884-886. | 6.1 | 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. | 1.5 | 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. | 1.1 | 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. | 1.4 | 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. | 1.1 | 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. | 1.1 | 9 |
| 121 | Individualized Iterative Phenotyping for Genome-wide Analysis of Loss-of-Function Mutations. American Journal of Human Genetics, 2015, 96, 913-925. | 2.6 | 66 |
| 122 | The somatic autosomal mutation matrix in cancer genomes. Human Genetics, 2015, 134, 851-864. | 1.8 | 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. | 6.5 | 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. | 1.4 | 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. | 1.8 | 52 |

| # | Article | IF | Citations |
|---|---|---|---|
| 127 | A Changing of the Guard at Human Genetics. Human Genetics, 2015, 134, 1-1. | 1.8 | 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. | 1.1 | 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. | 1.4 | 17 |
| 130 | An integrative approach to predicting the functional effects of non-coding and coding sequence variation. Bioinformatics, 2015, 31, 1536-1543. | 1.8 | 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. | 1.8 | 25 |
| 132 | Effect of predicted protein-truncating genetic variants on the human transcriptome. Science, 2015, 348, 666-669. | 6.0 | 252 |
| 133 | Mountain gorilla genomes reveal the impact of long-term population decline and inbreeding. Science, 2015, 348, 242-245. | 6.0 | 326 |
| 134 | A global reference for human genetic variation. Nature, 2015, 526, 68-74. | 13.7 | 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. | 3.3 | 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. | 1.4 | 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. | 1.4 | 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. | 1.4 | 40 |
| 139 | Mutations Causing Complex Disease May under Certain Circumstances Be Protective in an Epidemiological Sense. PLoS ONE, 2015, 10, e0132150. | 1.1 | 4 |
| 140 | Mechanisms of Base Substitution Mutagenesis in Cancer Genomes. Genes, 2014, 5, 108-146. | 1.0 | 49 |
| 141 | Key challenges for nextâ€generation pharmacogenomics. EMBO Reports, 2014, 15, 472-476. | 2.0 | 49 |
| 142 | A Probabilistic Model to Predict Clinical Phenotypic Traits from Genome Sequencing. PLoS Computational Biology, 2014, 10, e1003825. | 1.5 | 10 |
| 143 | A Massively Parallel Pipeline to Clone DNA Variants and Examine Molecular Phenotypes of Human Disease Mutations. PLoS Genetics, 2014, 10, e1004819. | 1.5 | 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. | 0.8 | 46 |

| # | Article | IF | Citations |
|---|---|---|---|
| 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. | 1.1 | 17 |
| 146 | MutPred Splice: machine learning-based prediction of exonic variants that disrupt splicing. Genome Biology, 2014, 15, R19. | 13.9 | 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. | 1.8 | 3 |
| 148 | MutationTaster2: mutation prediction for the deep-sequencing age. Nature Methods, 2014, 11, 361-362. | 9.0 | 3,203 |
| 149 | Impact of human pathogenic micro-insertions and micro-deletions on post-transcriptional regulation. Human Molecular Genetics, 2014, 23, 3024-3034. | 1.4 | 27 |
| 150 | Elucidating Common Structural Features of Human Pathogenic Variations Using Large-Scale Atomic-Resolution Protein Networks. Human Mutation, 2014, 35, 585-593. | 1.1 | 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. | 1.8 | 1,153 |
| 152 | Bridging genomics research between developed and developing countries: the Genomic Medicine Alliance. Personalized Medicine, 2014, 11, 615-623. | 0.8 | 22 |
| 153 | A New and More Accurate Estimate of the Rate of Concurrent Tandem-Base Substitution Mutations in the Human Germline: â^¼0.4% of the Single-Nucleotide Substitution Mutation Rate. Human Mutation, 2014, 35, 392-394. | 1.1 | 15 |
| 154 | Diagnostic Exome Sequencing to Elucidate the Genetic Basis of Likely Recessive Disorders in Consanguineous Families. Human Mutation, 2014, 35, 1203-1210. | 1.1 | 75 |
| 155 | Ranking non-synonymous single nucleotide polymorphisms based on disease concepts. Human Genomics, 2014, 8, 11. | 1.4 | 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. | 1.8 | 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. | 13.9 | 63 |
| 158 | Deciphering next-generation pharmacogenomics: an information technology perspective. Open Biology, 2014, 4, 140071. | 1.5 | 41 |
| 159 | Identification of LargeNF1Duplications Reciprocal to NAHR-Mediated Type-1NF1Deletions. Human Mutation, 2014, 35, 1469-1475. | 1.1 | 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. | 1.1 | 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. | 0.8 | 9 |
| 162 | New clinical and molecular insights on Barth syndrome. Orphanet Journal of Rare Diseases, 2013, 8, 27. | 1.2 | 35 |

| # | Article | IF | Citations |
|---|---|---|---|
| 163 | DDIG-in: discriminating between disease-associated and neutral non-frameshifting micro-indels. Genome Biology, 2013, 14, R23. | 13.9 | 63 |
| 164 | Identifying Mendelian disease genes with the Variant Effect Scoring Tool. BMC Genomics, 2013, 14, S3. | 1.2 | 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. | 1.8 | 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. | 1.8 | 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. | 1.4 | 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. | 1.4 | 4 |
| 173 | DNA structure matters. Genome Medicine, 2013, 5, 51. | 3.6 | 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. | 1.5 | 33 |
| 175 | Predicting the Functional, Molecular, and Phenotypic Consequences of Amino Acid Substitutions using Hidden Markov Models. Human Mutation, 2013, 34, 57-65. | 1.1 | 1,057 |
| 176 | Integrating nextâ€generation sequencing into the diagnostic testing of inherited cancer predisposition. Clinical Genetics, 2013, 83, 2-6. | 1.0 | 23 |
| 177 | Clinical relevance of cancer genome sequencing. World Journal of Gastroenterology, 2013, 19, 2011. | 1.4 | 16 |
| 178 | A new paradigm emerges from the study of de novo mutations in the context of neurodevelopmental disease. Molecular Psychiatry, 2013, 18, 141-153. | 4.1 | 85 |
| 179 | Patterns and Mutational Signatures of Tandem Base Substitutions Causing Human Inherited Disease. Human Mutation, 2013, 34, 1119-1130. | 1.1 | 34 |
| 180 | Predicting the functional consequences of cancer-associated amino acid substitutions. Bioinformatics, 2013, 29, 1504-1510. | 1.8 | 208 |

| # | Article | IF | Citations |
|---|---|---|---|
| 181 | CRAVAT: cancer-related analysis of variants toolkit. Bioinformatics, 2013, 29, 647-648. | 1.8 | 140 |
| 182 | Guanine Holes Are Prominent Targets for Mutation in Cancer and Inherited Disease. PLoS Genetics, 2013, 9, e1003816. | 1.5 | 34 |
| 183 | Research and clinical applications of cancer genome sequencing. Current Opinion in Obstetrics and Gynecology, 2013, 25, 3-10. | 0.9 | 14 |
| 184 | Using Exome Data to Identify Malignant Hyperthermia Susceptibility Mutations. Anesthesiology, 2013, 119, 1043-1053. | 1.3 | 69 |
| 185 | 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. | 1.1 | 12 |
| 186 | A Conservative Assessment of the Major Genetic Causes of Idiopathic Chronic Pancreatitis: Data from a Comprehensive Analysis of PRSS1, SPINK1, CTRC and CFTR Genes in 253 Young French Patients. PLoS ONE, 2013, 8, e73522. | 1.1 | 89 |
| 187 | Interlocus gene conversion events introduce deleterious mutations into at least 1% of human genes associated with inherited disease. Genome Research, 2012, 22, 429-435. | 2.4 | 30 |
| 188 | regSNPs: a strategy for prioritizing regulatory single nucleotide substitutions. Bioinformatics, 2012, 28, 1879-1886. | 1.8 | 13 |
| 189 | The â€˜sequence everythingâ€™ approach and personalized clinical decision challenges. Expert Review of Molecular Diagnostics, 2012, 12, 319-322. | 1.5 | 0 |
| 190 | Exploring the somatic NF1 mutational spectrum associated with NF1 cutaneous neurofibromas. European Journal of Human Genetics, 2012, 20, 411-419. | 1.4 | 25 |
| 191 | Ascertainment and critical assessment of the views of the general public and healthcare professionals on nutrigenomics in Greece. Personalized Medicine, 2012, 9, 201-210. | 0.8 | 20 |
| 192 | The Human Gene Mutation Database (HGMD) and Its Exploitation in the Fields of Personalized Genomics and Molecular Evolution. Current Protocols in Bioinformatics, 2012, 39, Unit1.13. | 25.8 | 198 |
| 193 | How to distinguish genetically between an alleged father and his monozygotic twin: A thought experiment. Forensic Science International: Genetics, 2012, 6, e129-e130. | 1.6 | 14 |
| 194 | Exome sequencing: a transient technology for molecular diagnostics?. Expert Review of Molecular Diagnostics, 2012, 12, 211-214. | 1.5 | 7 |
| 195 | Dissecting the clinical phenotype associated with mosaic type-2 NF1 microdeletions. Neurogenetics, 2012, 13, 229-236. | 0.7 | 20 |
| 196 | Exome versus transcriptome sequencing in identifying coding region variants. Expert Review of Molecular Diagnostics, 2012, 12, 241-251. | 1.5 | 43 |
| 197 | Assessment of the potential pathogenicity of missense mutations identified in the GTPase-activating protein (GAP)-related domain of the neurofibromatosis type-1 (NF1) gene. Human Mutation, 2012, 33, 1687-1696. | 1.1 | 21 |
| 198 | Identification of recurrent type-2NF1microdeletions reveals a mitotic nonallelic homologous recombination hotspot underlying a human genomic disorder. Human Mutation, 2012, 33, 1599-1609. | 1.1 | 26 |

| # | Article | IF | Citations |
|---|---|---|---|
| 199 | Non-coding RNA ANRIL and the number of plexiform neurofibromas in patients with NF1microdeletions. BMC Medical Genetics, 2012, 13, 98. | 2.1 | 11 |
| 200 | â€˜Sifting the significance from the dataâ€™ - the impact of high-throughput genomic technologies on human genetics and health care. Human Genomics, 2012, 6, 11. | 1.4 | 5 |
| 201 | Genotype-phenotype associations in neurofibromatosis type 1 (NF1): an increased risk of tumor complications in patients with NF1splice-site mutations?. Human Genomics, 2012, 6, 12. | 1.4 | 50 |
| 202 | Molecular heterogeneity in malignant peripheral nerve sheath tumors associated with neurofibromatosis type 1. Human Genomics, 2012, 6, 18. | 1.4 | 21 |
| 203 | An emerging role for microRNAs in NF1 tumorigenesis. Human Genomics, 2012, 6, 23. | 1.4 | 16 |
| 204 | A new era in the discovery of de novomutations underlying human genetic disease. Human Genomics, 2012, 6, 27. | 1.4 | 20 |
| 205 | Local sequence determinants of two in-frame triplet deletion/duplication hotspots in the RHD/RHCEgenes. Human Genomics, 2012, 6, 8. | 1.4 | 2 |
| 206 | An integrated map of genetic variation from 1,092 human genomes. Nature, 2012, 491, 56-65. | 13.7 | 7,199 |
| 207 | Gene discovery in familial cancer syndromes by exome sequencing: prospects for the elucidation of familial colorectal cancer type X. Modern Pathology, 2012, 25, 1055-1068. | 2.9 | 35 |
| 208 | Technological advances in DNA sequence enrichment and sequencing for germline genetic diagnosis. Expert Review of Molecular Diagnostics, 2012, 12, 159-173. | 1.5 | 16 |
| 209 | Deleterious- and Disease-Allele Prevalence in Healthy Individuals: Insights from Current Predictions, Mutation Databases, and Population-Scale Resequencing. American Journal of Human Genetics, 2012, 91, 1022-1032. | 2.6 | 255 |
| 210 | The Germline Mutational Spectrum in Neurofibromatosis Type 1 and Genotypeâ€“Phenotype Correlations. , 2012, , 115-134. | | 6 |
| 211 | Microattribution and nanopublication as means to incentivize the placement of human genome variation data into the public domain. Human Mutation, 2012, 33, 1503-1512. | 1.1 | 59 |
| 212 | A Systematic Survey of Loss-of-Function Variants in Human Protein-Coding Genes. Science, 2012, 335, 823-828. | 6.0 | 1,095 |
| 213 | The yak genome and adaptation to life at high altitude. Nature Genetics, 2012, 44, 946-949. | 9.4 | 708 |
| 214 | Insights into hominid evolution from the gorilla genome sequence. Nature, 2012, 483, 169-175. | 13.7 | 663 |
| 215 | Transient hypermutability, chromothripsis and replication-based mechanisms in the generation of concurrent clustered mutations. Mutation Research - Reviews in Mutation Research, 2012, 750, 52-59. | 2.4 | 25 |
| 216 | Characterization of the nonallelic homologous recombination hotspot PRS3 associated with type-3NF1deletions. Human Mutation, 2012, 33, 372-383. | 1.1 | 28 |

| # | Article | IF | Citations |
|---|---|---|---|
| 217 | Tissue-specific differences in the proportion of mosaic large NF1 deletions are suggestive of a selective growth advantage of hematopoietic del(+/â˜) stem cells. Human Mutation, 2012, 33, 541-550. | 1.1 | 23 |
| 218 | Microarray-based copy number analysis of neurofibromatosis type-1 (NF1)-associated malignant peripheral nerve sheath tumors reveals a role for Rho-GTPase pathway genes in NF1 tumorigenesis. Human Mutation, 2012, 33, 763-776. | 1.1 | 44 |
| 219 | Exome sequencing: Dual role as a discovery and diagnostic tool. Annals of Neurology, 2012, 71, 5-14. | 2.8 | 157 |
| 220 | NF1 Microdeletions and Their Underlying Mutational Mechanisms. , 2012, , 187-209. | | 10 |
| 221 | The Somatic Mutational Spectrum of the NF1 Gene. , 2012, , 211-233. | | 3 |
| 222 | Somatic Copy Number Alterations: Gene and Protein Expression Correlates in NF1-Associated Malignant Peripheral Nerve Sheath Tumors. , 2012, , 405-428. | | 0 |
| 223 | Genome sequencing and comparison of two nonhuman primate animal models, the cynomolgus and Chinese rhesus macaques. Nature Biotechnology, 2011, 29, 1019-1023. | 9.4 | 284 |
| 224 | The functional spectrum of low-frequency coding variation. Genome Biology, 2011, 12, R84. | 13.9 | 173 |
| 225 | Critical appraisal of the private genetic and pharmacogenomic testing environment in Greece. Personalized Medicine, 2011, 8, 413-420. | 0.8 | 20 |
| 226 | Exploring the potential relevance of human-specific genes to complex disease. Human Genomics, 2011, 5, 99. | 1.4 | 30 |
| 227 | A meta-analysis of single base-pair substitutions in translational termination codons ('nonstop') Tj ETQq1 1 0.784314 rgBT /Overlock | 1.4 | 34 |
| 228 | Cross-comparison of the genome sequences from human, chimpanzee, Neanderthal and a Denisovan hominin identifies novel potentially compensated mutations. Human Genomics, 2011, 5, 453. | 1.4 | 3 |
| 229 | Human genetics and genomics a decade after the release of the draft sequence of the human genome. Human Genomics, 2011, 5, 577. | 1.4 | 86 |
| 230 | Neurofibromatosis type 1-associated tumours: Their somatic mutational spectrum and pathogenesis. Human Genomics, 2011, 5, 623. | 1.4 | 113 |
| 231 | Local DNA sequence determinants of FUT2 copy number variation. Transfusion, 2011, 51, 1359-1361. | 0.8 | 3 |
| 232 | Assessing the pathological relevance of SPINK1 promoter variants. European Journal of Human Genetics, 2011, 19, 1066-1073. | 1.4 | 18 |
| 233 | Mapping copy number variation by population-scale genome sequencing. Nature, 2011, 470, 59-65. | 13.7 | 991 |
| 234 | Variation in genome-wide mutation rates within and between human families. Nature Genetics, 2011, 43, 712-714. | 9.4 | 525 |

| # | Article | IF | Citations |
|---|---|---|---|
| 235 | Delineating the Hemostaseome as an aid to individualize the analysis of the hereditary basis of thrombotic and bleeding disorders. Human Genetics, 2011, 130, 149-166. | 1.8 | 12 |
| 236 | Lionizing lyonization 50Âyears on. Human Genetics, 2011, 130, 167-168. | 1.8 | 4 |
| 237 | 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. | 1.2 | 32 |
| 238 | Delineation of the clinical phenotype associated with non-mosaic type-2 NF1 deletions: two case reports. Journal of Medical Case Reports, 2011, 5, 577. | 0.4 | 12 |
| 239 | Meiotic recombination favors the spreading of deleterious mutations in human populations. Human Mutation, 2011, 32, 198-206. | 1.1 | 37 |
| 240 | Mosaic type-1 NF1 microdeletions as a cause of both generalized and segmental neurofibromatosis type-1 (NF1). Human Mutation, 2011, 32, 213-219. | 1.1 | 106 |
| 241 | Monozygotic twins discordant for neurofibromatosis type 1 due to a postzygotic NF1 gene mutation. Human Mutation, 2011, 32, E2134-E2147. | 1.1 | 34 |
| 242 | IDUA mutational profiling of a cohort of 102 European patients with mucopolysaccharidosis type I: identification and characterization of 35 novel Î±-L-iduronidase (IDUA) alleles. Human Mutation, 2011, 32, E2189-E2210. | 1.1 | 66 |
| 243 | Comparative analysis of germline and somatic microlesion mutational spectra in 17 human tumor suppressor genes. Human Mutation, 2011, 32, 620-632. | 1.1 | 13 |
| 244 | Single base-pair substitutions at the translation initiation sites of human genes as a cause of inherited disease. Human Mutation, 2011, 32, 1137-1143. | 1.1 | 32 |
| 245 | On the sequence-directed nature of human gene mutation: The role of genomic architecture and the local DNA sequence environment in mediating gene mutations underlying human inherited disease. Human Mutation, 2011, 32, 1075-1099. | 1.1 | 99 |
| 246 | Prediction of functional regulatory SNPs in monogenic and complex disease. Human Mutation, 2011, 32, 1183-1190. | 1.1 | 21 |
| 247 | Demographic history and rare allele sharing among human populations. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 11983-11988. | 3.3 | 589 |
| 248 | A critical view of the general publicâ€™s awareness and physiciansâ€™ opinion of the trends and potential pitfalls of genetic testing in Greece. Personalized Medicine, 2011, 8, 551-561. | 0.8 | 47 |
| 249 | Non-B DNA-forming Sequences and WRN Deficiency Independently Increase the Frequency of Base Substitution in Human Cells. Journal of Biological Chemistry, 2011, 286, 10017-10026. | 1.6 | 31 |
| 250 | Loss of exon identity is a common mechanism of human inherited disease. Genome Research, 2011, 21, 1563-1571. | 2.4 | 156 |
| 251 | Functional intronic polymorphisms: Buried treasure awaiting discovery within our genes. Human Genomics, 2010, 4, 284. | 1.4 | 192 |
| 252 | 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. | 1.4 | 10 |

| # | Article | IF | Citations |
|---|---|---|---|
| 253 | Methylation-mediated deamination of 5-methylcytosine appears to give rise to mutations causing human inherited disease in CpNpG trinucleotides, as well as in CpG dinucleotides. Human Genomics, 2010, 4, 406. | 1.4 | 118 |
| 254 | Prospects for the automated extraction of mutation data from the scientific literature. Human Genomics, 2010, 5, 1. | 1.4 | 4 |
| 255 | Characterisation of a functional intronic polymorphism in the human growth hormone (GHI) gene. Human Genomics, 2010, 4, 289. | 1.4 | 41 |
| 256 | Genomic rearrangements in inherited disease and cancer. Seminars in Cancer Biology, 2010, 20, 222-233. | 4.3 | 140 |
| 257 | Structure-based kernels for the prediction of catalytic residues and their involvement in human inherited disease. BMC Bioinformatics, 2010, 11, . | 1.2 | 6 |
| 258 | Extended runs of homozygosity at 17q11.2: an association with type-2NF1deletions?. Human Mutation, 2010, 31, 325-334. | 1.1 | 9 |
| 259 | In silico functional profiling of human disease-associated and polymorphic amino acid substitutions. Human Mutation, 2010, 31, 335-346. | 1.1 | 57 |
| 260 | Complete ascertainment of intragenic copy number mutations (CNMs) in the CFTR gene and its implications for CNM formation at other autosomal loci. Human Mutation, 2010, 31, 421-428. | 1.1 | 31 |
| 261 | Enigmatic In Vivo iduronate-2-sulfatase (IDS) mutant transcript correction to wild-type in Hunter syndrome. Human Mutation, 2010, 31, E1261-E1285. | 1.1 | 17 |
| 262 | A novel third type of recurrent NF1 microdeletion mediated by nonallelic homologous recombination between LRRC37B-containing low-copy repeats in 17q11.2. Human Mutation, 2010, 31, 742-751. | 1.1 | 42 |
| 263 | Genes, mutations, and human inherited disease at the dawn of the age of personalized genomics. Human Mutation, 2010, 31, 631-655. | 1.1 | 161 |
| 264 | Intrachromosomal mitotic nonallelic homologous recombination is the major molecular mechanism underlying type-2 NF1 deletions. Human Mutation, 2010, 31, 1163-1173. | 1.1 | 36 |
| 265 | Identification and characterization of 15 novel GALC gene mutations causing Krabbe disease. Human Mutation, 2010, 31, E1894-E1914. | 1.1 | 93 |
| 266 | Triangulation of the human, chimpanzee, and Neanderthal genome sequences identifies potentially compensated mutations. Human Mutation, 2010, 31, 1286-1293. | 1.1 | 12 |
| 267 | Is the NIH policy for sharing GWAS data running the risk of being counterproductive?. Investigative Genetics, 2010, 1, 3. | 3.3 | 4 |
| 268 | Legal and ethical consequences of international biobanking from a national perspective: the German BMB-EUCoop project. European Journal of Human Genetics, 2010, 18, 522-525. | 1.4 | 16 |
| 269 | GWAS: heritability missing in action?. European Journal of Human Genetics, 2010, 18, 859-861. | 1.4 | 74 |
| 270 | A map of human genome variation from population-scale sequencing. Nature, 2010, 467, 1061-1073. | 13.7 | 7,209 |

| # | Article | IF | Citations |
|---|---|---|---|
| 271 | Revealing the human mutome. Clinical Genetics, 2010, 78, 310-320. | 1.0 | 22 |
| 272 | Gene Conversion in Human Genetic Disease. Genes, 2010, 1, 550-563. | 1.0 | 16 |
| 273 | Structure-based kernels for the prediction of catalytic residues and their involvement in human inherited disease. Bioinformatics, 2010, 26, 1975-1982. | 1.8 | 15 |
| 274 | Do Inherited Disease Genes Have Distinguishing Functional Characteristics?. Genetic Testing and Molecular Biomarkers, 2010, 14, 289-291. | 0.3 | 4 |
| 275 | Human Gene Mutation: Mechanisms and Consequences. , 2010, , 319-363. | | 6 |
| 276 | Clinical characterisation of 29 neurofibromatosis type-1 patients with molecularly ascertained 1.4 Mb type-1 NF1 deletions. Journal of Medical Genetics, 2010, 47, 623-630. | 1.5 | 148 |
| 277 | Chromosomal Distribution of Disease Genes in the Human Genome. Genetic Testing and Molecular Biomarkers, 2010, 14, 441-446. | 0.3 | 3 |
| 278 | Splicing factor SFRS1 recognizes a functionally diverse landscape of RNA transcripts. Genome Research, 2009, 19, 381-394. | 2.4 | 284 |
| 279 | Elucidation of the complex structure and origin of the human trypsinogen locus triplication. Human Molecular Genetics, 2009, 18, 3605-3614. | 1.4 | 22 |
| 280 | Automated inference of molecular mechanisms of disease from amino acid substitutions. Bioinformatics, 2009, 25, 2744-2750. | 1.8 | 691 |
| 281 | Gene synteny comparisons between different vertebrates provide new insights into breakage and fusion events during mammalian karyotype evolution. BMC Evolutionary Biology, 2009, 9, 84. | 3.2 | 54 |
| 282 | A gene conversion hotspot in the human growth hormone (GH1) gene promoter. Human Mutation, 2009, 30, 239-247. | 1.1 | 13 |
| 283 | Identification and molecular characterization of six novel mutations in the UDP-N-acetylglucosamine-1-phosphotransferase gamma subunit (GNPTG) gene in patients with mucolipidosis III gamma. Human Mutation, 2009, 30, 978-984. | 1.1 | 26 |
| 284 | Gene conversion causing human inherited disease: Evidence for involvement of non-B-DNA-forming sequences and recombination-promoting motifs in DNA breakage and repair. Human Mutation, 2009, 30, 1189-1198. | 1.1 | 63 |
| 285 | Closely spaced multiple mutations as potential signatures of transient hypermutability in human genes. Human Mutation, 2009, 30, 1435-1448. | 1.1 | 51 |
| 286 | Molecular characterization of 22 novel UDP-N-acetylglucosamine-1-phosphate transferase Î±- and Î²-subunit (GNPTAB) gene mutations causing mucolipidosis types IIÎ±/Î² and IIIÎ±/Î² in 46 patients. Human Mutation, 2009, 30, E956-E973. | 1.1 | 38 |
| 287 | Air pollution and mutations in the germline: are humans at risk?. Human Genetics, 2009, 125, 119-130. | 1.8 | 20 |
| 288 | STREGA: a â€˜How-Toâ€™ guide for reporting genetic associations. Human Genetics, 2009, 125, 117-118. | 1.8 | 5 |

| # | Article | IF | Citations |
|---|---|---|---|
| 289 | Cruciform-forming inverted repeats appear to have mediated many of the microinversions that distinguish the human and chimpanzee genomes. Chromosome Research, 2009, 17, 469-483. | 1.0 | 31 |
| 290 | Mechanisms of Loss of Heterozygosity in Neurofibromatosis Type 1-Associated Plexiform Neurofibromas. Journal of Investigative Dermatology, 2009, 129, 615-621. | 0.3 | 42 |
| 291 | The Human Gene Mutation Database: 2008 update. Genome Medicine, 2009, 1, 13. | 3.6 | 774 |
| 292 | The Human Gene Mutation Database: providing a comprehensive central mutation database for molecular diagnostics and personalised genomics. Human Genomics, 2009, 4, 69. | 1.4 | 151 |
| 293 | Molecular mechanisms of chromosomal rearrangement during primate evolution. Chromosome Research, 2008, 16, 41-56. | 1.0 | 68 |
| 294 | A meta-analysis of nonsense mutations causing human genetic disease. Human Mutation, 2008, 29, 1037-1047. | 1.1 | 348 |
| 295 | Two sisters with Rett syndrome and non-identical paternally-derived microdeletions in the MECP2 gene. Genomic Medicine, 2008, 2, 77-81. | 0.6 | 4 |
| 296 | Copy number variations in the NF1 gene region are infrequent and do not predispose to recurrent type-1 deletions. European Journal of Human Genetics, 2008, 16, 572-580. | 1.4 | 22 |
| 297 | Detection of two Alu insertions in the CFTR gene. Journal of Cystic Fibrosis, 2008, 7, 37-43. | 0.3 | 38 |
| 298 | 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. | 1.6 | 16 |
| 299 | Mosaicism in sporadic neurofibromatosis type 1: variations on a theme common to other hereditary cancer syndromes?. Journal of Medical Genetics, 2008, 45, 622-631. | 1.5 | 71 |
| 300 | Comparative analysis of copy number variation in primate genomes. Cytogenetic and Genome Research, 2008, 123, 288-296. | 0.6 | 5 |
| 301 | Preface. Cytogenetic and Genome Research, 2008, 123, 5-6. | 0.6 | 2 |
| 302 | Abundance and length of simple repeats in vertebrate genomes are determined by their structural properties. Genome Research, 2008, 18, 1545-1553. | 2.4 | 87 |
| 303 | Human Gene Mutation Database: towards a comprehensive central mutation database. Journal of Medical Genetics, 2007, 45, 124-126. | 1.5 | 90 |
| 304 | Chromosomal speciation of humans and chimpanzees revisited: studies of DNA divergence within inverted regions. Cytogenetic and Genome Research, 2007, 116, 53-60. | 0.6 | 13 |
| 305 | Co-inheritance of a novel deletion of the entire SPINK1 gene with a CFTR missense mutation (L997F) in a family with chronic pancreatitis. Molecular Genetics and Metabolism, 2007, 92, 168-175. | 0.5 | 25 |
| 306 | Gain-of-glycosylation mutations. Current Opinion in Genetics and Development, 2007, 17, 245-251. | 1.5 | 65 |

| # | Article | IF | Citations |
|---|---|---|---|
| 307 | Type 2 NF1 Deletions Are Highly Unusual by Virtue of the Absence of Nonallelic Homologous Recombination Hotspots and an Apparent Preference for Female Mitotic Recombination. American Journal of Human Genetics, 2007, 81, 1201-1220. | 2.6 | 60 |
| 308 | Evolutionary and Biomedical Insights from the Rhesus Macaque Genome. Science, 2007, 316, 222-234. | 6.0 | 1,283 |
| 309 | Single base-pair substitutions in exon-intron junctions of human genes: nature, distribution, and consequences for mRNA splicing. Human Mutation, 2007, 28, 150-158. | 1.1 | 324 |
| 310 | Understanding the recent evolution of the human genome: insights from human-chimpanzee genome comparisons. Human Mutation, 2007, 28, 99-130. | 1.1 | 98 |
| 311 | Diversity of cystathionine Î²-synthase haplotypes bearing the most common homocystinuria mutation c.833T>C: a possible role for gene conversion. Human Mutation, 2007, 28, 255-264. | 1.1 | 20 |
| 312 | Gene conversion: mechanisms, evolution and human disease. Nature Reviews Genetics, 2007, 8, 762-775. | 7.7 | 576 |
| 313 | A legal framework for biobanking: the German experience. European Journal of Human Genetics, 2007, 15, 528-532. | 1.4 | 17 |
| 314 | Compound heterozygosity for two novel mutations (1203insG/Y1456X) in the von Willebrand factor gene causing type 3 von Willebrand disease. Haemophilia, 2007, 13, 645-648. | 1.0 | 4 |
| 315 | Structural divergence between the human and chimpanzee genomes. Human Genetics, 2007, 120, 759-778. | 1.8 | 49 |
| 316 | Searching for potential microRNA-binding site mutations amongst known disease-associated 3â€² UTR variants. Genomic Medicine, 2007, 1, 29-33. | 0.6 | 7 |
| 317 | Mechanism of Alu integration into the human genome. Genomic Medicine, 2007, 1, 9-17. | 0.6 | 16 |
| 318 | Molecular cytogenetic characterization of two independent karyotypic anomalies in a patient with severe mental retardation and juvenile idiopathic arthritis. Genomic Medicine, 2007, 1, 65-73. | 0.6 | 0 |
| 319 | A novel Alu-mediated 61-kb deletion of the von Willebrand factor (VWF) gene whose breakpoints co-locate with putative matrix attachment regions. Blood Cells, Molecules, and Diseases, 2006, 36, 385-391. | 0.6 | 19 |
| 320 | The chimpanzee-specific pericentric inversions that distinguish humans and chimpanzees have identical breakpoints in Pan troglodytes and Pan paniscus. Genomics, 2006, 87, 39-45. | 1.3 | 20 |
| 321 | Assessing Radiation-Associated Mutational Risk to the Germline: Repetitive DNA Sequences as Mutational Targets and Biomarkers. Radiation Research, 2006, 165, 249-268. | 0.7 | 45 |
| 322 | Utilization of a cryptic noncanonical donor splice site in the KRT14 gene causes a mild form of epidermolysis bullosa simplex. British Journal of Dermatology, 2006, 155, 201-203. | 1.4 | 7 |
| 323 | Gross genomic rearrangements involving deletions in the CFTR gene: characterization of six new events from a large cohort of hitherto unidentified cystic fibrosis chromosomes and meta-analysis of the underlying mechanisms. European Journal of Human Genetics, 2006, 14, 567-576. | 1.4 | 77 |
| 324 | Polymorphic micro-inversions contribute to the genomic variability of humans and chimpanzees. Human Genetics, 2006, 119, 103-112. | 1.8 | 17 |

| # | Article | IF | Citations |
|---|---|---|---|
| 325 | Identification of large-scale human-specific copy number differences by inter-species array comparative genomic hybridization. Human Genetics, 2006, 119, 185-198. | 1.8 | 35 |
| 326 | Genetic variation at the growth hormone (GH1) and growth hormone receptor (GHR) loci as a risk factor for hypertension and stroke. Human Genetics, 2006, 119, 527-540. | 1.8 | 29 |
| 327 | A systematic analysis of disease-associated variants in the 3â€² regulatory regions of human protein-coding genes I: general principles and overview. Human Genetics, 2006, 120, 1-21. | 1.8 | 135 |
| 328 | Characterization of the human lineage-specific pericentric inversion that distinguishes human chromosome 1 from the homologous chromosomes of the great apes. Human Genetics, 2006, 120, 126-138. | 1.8 | 17 |
| 329 | Complex patterns of copy number variation at sites of segmental duplications: an important category of structural variation in the human genome. Human Genetics, 2006, 120, 270-284. | 1.8 | 68 |
| 330 | A systematic analysis of disease-associated variants in the 3â€² regulatory regions of human protein-coding genes II: the importance of mRNA secondary structure in assessing the functionality of 3â€² UTR variants. Human Genetics, 2006, 120, 301-333. | 1.8 | 125 |
| 331 | Origin of the prevalentSFTPBindel g.1549Câ€‰›â€‰GAA (121ins2) mutation causing surfactant protein B (SP-B) deficiency. American Journal of Medical Genetics, Part A, 2006, 140A, 62-69. | 0.7 | 23 |
| 332 | Long homopurine*homopyrimidine sequences are characteristic of genes expressed in brain and the pseudoautosomal region. Nucleic Acids Research, 2006, 34, 2663-2675. | 6.5 | 60 |
| 333 | LINE-1 Endonuclease-Dependent Retrotranspositional Events Causing Human Genetic Disease: Mutation Detection Bias and Multiple Mechanisms of Target Gene Disruption. Journal of Biomedicine and Biotechnology, 2006, 2006, 1-9. | 3.0 | 51 |
| 334 | In silico discrimination of single nucleotide polymorphisms and pathological mutations in human gene promoter regions by means of local DNA sequence context and regularity. In Silico Biology, 2006, 6, 23-34. | 0.4 | 15 |
| 335 | The Human Gene Mutation Database (HGMD) and Its Exploitation in the Study of Mutational Mechanisms. Current Protocols in Bioinformatics, 2005, 12, 1.13.1-1.13.20. | 25.8 | 50 |
| 336 | Metaâ€Analysis of gross insertions causing human genetic disease: Novel mutational mechanisms and the role of replication slippage. Human Mutation, 2005, 25, 207-221. | 1.1 | 148 |
| 337 | Gains of glycosylation comprise an unexpectedly large group of pathogenic mutations. Nature Genetics, 2005, 37, 692-700. | 9.4 | 198 |
| 338 | Early onset seizures and Rett-like features associated with mutations in CDKL5. European Journal of Human Genetics, 2005, 13, 1113-1120. | 1.4 | 160 |
| 339 | Gonosomal Mosaicism for a Nonsense Mutation (R1947X) in the NF1 Gene in Segmental Neurofibromatosis Type 1. Journal of Investigative Dermatology, 2005, 125, 463-466. | 0.3 | 68 |
| 340 | Breakpoint analysis of the pericentric inversion distinguishing human chromosome 4 from the homologous chromosome in the chimpanzee (Pan troglodytes). Human Mutation, 2005, 25, 45-55. | 1.1 | 47 |
| 341 | Complex gene rearrangements caused by serial replication slippage. Human Mutation, 2005, 26, 125-134. | 1.1 | 88 |
| 342 | Microdeletions and microinsertions causing human genetic disease: common mechanisms of mutagenesis and the role of local DNA sequence complexity. Human Mutation, 2005, 26, 205-213. | 1.1 | 136 |

| # | Article | IF | Citations |
|---|---|---|---|
| 343 | Intrachromosomal serial replication slippage intransgives rise to diverse genomic rearrangements involving inversions. Human Mutation, 2005, 26, 362-373. | 1.1 | 62 |
| 344 | Molecular characterisation of the pericentric inversion that distinguishes human chromosome 5 from the homologous chimpanzee chromosome. Human Genetics, 2005, 117, 168-176. | 1.8 | 27 |
| 345 | A systematic analysis of LINE-1 endonuclease-dependent retrotranspositional events causing human genetic disease. Human Genetics, 2005, 117, 411-427. | 1.8 | 206 |
| 346 | Independent intrachromosomal recombination events underlie the pericentric inversions of chimpanzee and gorilla chromosomes homologous to human chromosome 16. Genome Research, 2005, 15, 1232-1242. | 2.4 | 42 |
| 347 | Gross rearrangements of the MECP2 gene are found in both classical and atypical Rett syndrome patients. Journal of Medical Genetics, 2005, 43, 451-456. | 1.5 | 62 |
| 348 | Neurofibromatosis Type 1: A Common Familial Cancer Syndrome. , 2004, , 285-310. | | 2 |
| 349 | A Novel Dysfunctional Growth Hormone Variant (Ile179Met) Exhibits a Decreased Ability to Activate the Extracellular Signal-Regulated Kinase Pathway. Journal of Clinical Endocrinology and Metabolism, 2004, 89, 1068-1075. | 1.8 | 53 |
| 350 | Breakpoints of gross deletions coincide with non-B DNA conformations. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 14162-14167. | 3.3 | 184 |
| 351 | Genome sequence of the Brown Norway rat yields insights into mammalian evolution. Nature, 2004, 428, 493-521. | 13.7 | 1,943 |
| 352 | Functional analysis of polymorphic variation within the promoter and 5? untranslated region of the neurofibromatosis type 1 (NF1) gene. American Journal of Medical Genetics Part A, 2004, 131A, 227-231. | 2.4 | 9 |
| 353 | Characterization of the somatic mutational spectrum of the neurofibromatosis type 1 (NF1) gene in neurofibromatosis patients with benign and malignant tumors. Human Mutation, 2004, 23, 134-146. | 1.1 | 97 |
| 354 | Gross rearrangement breakpoint database (GRaBD?). Human Mutation, 2004, 23, 219-221. | 1.1 | 24 |
| 355 | Genomic rearrangements in theCFTRgene: Extensive allelic heterogeneity and diverse mutational mechanisms. Human Mutation, 2004, 23, 343-357. | 1.1 | 115 |
| 356 | Evolutionary conservation and selection of human disease gene orthologs in the rat and mouse genomes. Genome Biology, 2004, 5, R47. | 13.9 | 116 |
| 357 | Detection of NF1 Mutations Utilizing the Protein Truncation Test (PTT). , 2003, 217, 315-328. | | 3 |
| 358 | Three different pathological lesions in the NF1 gene originating de novo in a family with neurofibromatosis type 1. Human Genetics, 2003, 112, 12-17. | 1.8 | 24 |
| 359 | Human genetic disease caused by de novo mitochondrial-nuclear DNA transfer. Human Genetics, 2003, 112, 303-309. | 1.8 | 114 |
| 360 | Meta-analysis of indels causing human genetic disease: mechanisms of mutagenesis and the role of local DNA sequence complexity. Human Mutation, 2003, 21, 28-44. | 1.1 | 112 |

| # | Article | IF | Citations |
|---|---|---|---|
| 361 | Human growth hormone 1 (GH1) gene expression: Complex haplotype-dependent influence of polymorphic variation in the proximal promoter and locus control region. Human Mutation, 2003, 21, 408-423. | 1.1 | 99 |
| 362 | Novel mutations of the growth hormone 1 (GH1) gene disclosed by modulation of the clinical selection criteria for individuals with short stature. Human Mutation, 2003, 21, 424-440. | 1.1 | 106 |
| 363 | Human Gene Mutation Database (HGMDÂ®): 2003 update. Human Mutation, 2003, 21, 577-581. | 1.1 | 1,571 |
| 364 | Translocation and gross deletion breakpoints in human inherited disease and cancer II: Potential involvement of repetitive sequence elements in secondary structure formation between DNA ends. Human Mutation, 2003, 22, 245-251. | 1.1 | 98 |
| 365 | Translocation and gross deletion breakpoints in human inherited disease and cancer I: Nucleotide composition and recombination-associated motifs. Human Mutation, 2003, 22, 229-244. | 1.1 | 214 |
| 366 | Prenatal Exclusion of Severe Factor VII Deficiency. Journal of Pediatric Hematology/Oncology, 2003, 25, 418-420. | 0.3 | 13 |
| 367 | A rare complex DNA rearrangement in the murine Steel gene results in exon duplication and a lethal phenotype. Blood, 2003, 102, 3548-3555. | 0.6 | 5 |
| 368 | THE EVOLUTION OF THE VERTEBRATE Î²-GLOBIN GENE PROMOTER. Evolution; International Journal of Organic Evolution, 2002, 56, 224. | 1.1 | 2 |
| 369 | Molecular diagnosis of facioscapulohumeral muscular dystrophy. Expert Review of Molecular Diagnostics, 2002, 2, 160-171. | 1.5 | 28 |
| 370 | Assessing the relative importance of the biophysical properties of amino acid substitutions associated with human genetic disease. Human Mutation, 2002, 20, 98-109. | 1.1 | 39 |
| 371 | Proposed guidelines for papers describing DNA polymorphism-disease associations. Human Genetics, 2002, 110, 207-208. | 1.8 | 114 |
| 372 | THE EVOLUTION OF THE VERTEBRATE ?-GLOBIN GENE PROMOTER. Evolution; International Journal of Organic Evolution, 2002, 56, 224-232. | 1.1 | 7 |
| 373 | Human gene mutation in pathology and evolution. Journal of Inherited Metabolic Disease, 2002, 25, 157-182. | 1.7 | 38 |
| 374 | Human growth hormone I gene expression is influenced in a complex haplotype-dependent fashion by polymorphic variation in both the proximal promoter and the locus control region. Journal of Pediatric Endocrinology and Metabolism, 2002, 15 Suppl 5, 1429. | 0.4 | 0 |
| 375 | Estimating the Efficacy and Efficiency of Cascade Genetic Screening. American Journal of Human Genetics, 2001, 69, 361-370. | 2.6 | 62 |
| 376 | The Frequency of Inherited Disorders Database. Human Genetics, 2001, 108, 72-74. | 1.8 | 6 |
| 377 | Human type I hair keratin pseudogene ? hHaA has functional orthologs in the chimpanzee and gorilla: evidence for recent inactivation of the human gene after the Pan-Homo divergence. Human Genetics, 2001, 108, 37-42. | 1.8 | 87 |
| 378 | Evaluation of denaturing high performance liquid chromatography (DHPLC) for the mutational analysis of the neurofibromatosis type 1 ( NF1 ) gene. Human Genetics, 2001, 109, 487-497. | 1.8 | 71 |

| # | Article | IF | Citations |
|---|---|---|---|
| 379 | Molecular analysis of the 5â€²-flanking region of the neurofibromatosis type 1 (NF1) gene: identification of five sequence variants. Clinical Genetics, 2001, 57, 221-224. | 1.0 | 12 |
| 380 | 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. | 1.0 | 15 |
| 381 | Human Gene Mutation Database?A biomedical information and research resource. Human Mutation, 2000, 15, 45-51. | 1.1 | 241 |
| 382 | Identification of an intronic regulatory element in the human protein C ( PROC) gene. Human Genetics, 2000, 107, 458-465. | 1.8 | 20 |
| 383 | 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. | 1.8 | 13 |
| 384 | Molecular genetic analysis of severe protein C deficiency. Human Genetics, 2000, 106, 646-653. | 1.8 | 13 |
| 385 | Molecular analysis of the genotype-phenotype relationship in factor X deficiency. Human Genetics, 2000, 106, 249-257. | 1.8 | 71 |
| 386 | Long-read sequence analysis of the MECP2 gene in Rett syndrome patients: correlation of disease severity with mutation type and location. Human Molecular Genetics, 2000, 9, 1119-1129. | 1.4 | 245 |
| 387 | Promoter shuffling has occurred during the evolution of the vertebrate growth hormone gene. Gene, 2000, 254, 9-18. | 1.0 | 19 |
| 388 | Molecular genetic analysis of severe proteinÂC deficiency. Human Genetics, 2000, 106, 646-653. | 1.8 | 37 |
| 389 | 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. | 1.8 | 30 |
| 390 | Disease-causing mutations in the human genome. European Journal of Pediatrics, 2000, 159, S173-S178. | 1.3 | 64 |
| 391 | Disentangling the perturbational effects of amino acid substitutions in the DNA-binding domain of p53. Human Genetics, 1999, 104, 15-22. | 1.8 | 16 |
| 392 | Evolution of the proximal promoter region of the mammalian growth hormone gene. Gene, 1999, 237, 143-151. | 1.0 | 34 |
| 393 | Gross deletions of the neurofibromatosis type 1 (NF1) gene are predominantly of maternal origin and commonly associated with a learning disability, dysmorphic features and developmental delay. Human Genetics, 1998, 102, 591-597. | 1.8 | 171 |
| 394 | Variation of site-specific methylation patterns in the factor VIII ( F8C ) gene in human sperm DNA. Human Genetics, 1998, 103, 228-233. | 1.8 | 7 |
| 395 | The molecular genetics of growth hormone deficiency. Human Genetics, 1998, 103, 255-272. | 1.8 | 148 |
| 396 | Neighboring-Nucleotide Effects on the Rates of Germ-Line Single-Base-Pair Substitution in Human Genes. American Journal of Human Genetics, 1998, 63, 474-488. | 2.6 | 291 |

| # | Article | IF | Citations |
|---|---|---|---|
| 397 | The human gene mutation database. Nucleic Acids Research, 1998, 26, 285-287. | 6.5 | 231 |
| 398 | p53 mutations, benzo[a]pyrene and lung cancer. Mutagenesis, 1998, 13, 319-320. | 1.0 | 26 |
| 399 | The human gene mutation database. Trends in Genetics, 1997, 13, 121-122. | 2.9 | 249 |
| 400 | Homology modelling of the catalytic domain of early mammalian protein C: evolution of structural features. Human Genetics, 1997, 101, 37-42. | 1.8 | 5 |
| 401 | Inherited Factor VII Deficiency: Molecular Genetics and Pathophysiology. Thrombosis and Haemostasis, 1997, 78, 151-160. | 1.8 | 103 |
| 402 | Inherited Factor X Deficiency: Molecular Genetics and Pathophysiology. Thrombosis and Haemostasis, 1997, 78, 161-172. | 1.8 | 71 |
| 403 | [14]Analysis of promoter mutations causing human genetic disease. Methods in Molecular Genetics, 1996, 8, 261-277. | 0.6 | 0 |
| 404 | Molecular reconstruction and homology modelling of the catalytic domain of the common ancestor of the haemostatic vitamin-K-dependent serine proteinases. Human Genetics, 1996, 98, 351-370. | 1.8 | 14 |
| 405 | Mutational and functional analysis of the neurofibromatosis type 1 ( NF1 ) gene. Human Genetics, 1996, 99, 88-92. | 1.8 | 105 |
| 406 | Single base-pair substitutions in pathology and evolution: Two sides to the same coin. Human Mutation, 1996, 8, 23-31. | 1.1 | 23 |
| 407 | 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. | 1.8 | 38 |
| 408 | 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. | 1.8 | 7 |
| 409 | Somatic spectrum of cancer-associated single basepair substitutions in the TP53 gene is determined mainly by endogenous mechanisms of mutation and by selection. Human Mutation, 1995, 5, 48-57. | 1.1 | 56 |
| 410 | Prothrombin cleavage by human vascular smooth muscle cells: A potential alternative pathway to the coagulation cascade. Journal of Cellular Biochemistry, 1995, 59, 514-528. | 1.2 | 8 |
| 411 | The mutational demography of protein C deficiency. Human Genetics, 1995, 96, 142-146. | 1.8 | 10 |
| 412 | 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. | 1.8 | 2 |
| 413 | Core database. Nature, 1995, 374, 402-402. | 13.7 | 8 |
| 414 | Population differences in the frequency of the factor V Leiden variant among people with clinically symptomatic protein C deficiency.. Journal of Medical Genetics, 1995, 32, 543-545. | 1.5 | 25 |

| # | Article | IF | Citations |
|---|---|---|---|
| 415 | Haemophilia A: database of nucleotide substitutions, deletions, insertions and rearrangements of the factor VIII gene, second edition. Nucleic Acids Research, 1994, 22, 3511-3533. | 6.5 | 112 |
| 416 | Haemophilia A: database of nucleotide substitutions, deletions, insertions and rearrangements of the factor VIII gene, second edition. Nucleic Acids Research, 1994, 22, 4851-4868. | 6.5 | 56 |
| 417 | Disruption of a binding site for hepatocyte nuclear factor 1 in the protein C gene promoter is associated with hereditary thrombophilia. Human Molecular Genetics, 1994, 3, 2147-2152. | 1.4 | 29 |
| 418 | 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. | 1.8 | 20 |
| 419 | 11 The molecular genetics of familial venous thrombosis. Best Practice and Research: Clinical Haematology, 1994, 7, 637-674. | 1.1 | 12 |
| 420 | Ectopic (Illegitimate) Transcription: New Possibilities for the Analysis and Diagnosis of Human Genetic Disease. Annals of Medicine, 1994, 26, 9-14. | 1.5 | 41 |
| 421 | Screening for mutations in the antithrombin III gene causing recurrent venous thrombosis by single-strand conformation polymorphism analysis. Human Mutation, 1993, 2, 324-326. | 1.1 | 14 |
| 422 | Human Gene Mutations Affecting RNA Processing and Translation. Annals of Medicine, 1993, 25, 11-17. | 1.5 | 116 |
| 423 | Detection of missense mutations by single-strand conformational polymorphism (SSCP) analysis in five dysfunctional variants of coagulation factor VII. Human Molecular Genetics, 1993, 2, 1355-1359. | 1.4 | 30 |
| 424 | Regulatory Mutations and Human Genetic Disease. Annals of Medicine, 1992, 24, 427-437. | 1.5 | 26 |
| 425 | Molecular Genetic Approaches to the Analysis and Diagnosis of Human Inherited Disease: An Overview. Annals of Medicine, 1992, 24, 29-42. | 1.5 | 17 |
| 426 | De novo splice site mutation in the antithrombin III (AT3) gene causing recurrent venous thrombosis: Demonstration of exon skipping by ectopic transcript analysis. Genomics, 1992, 13, 1359-1361. | 1.3 | 33 |
| 427 | Single-strand conformation polymorphism (SSCP) analysis of exon 11 of the CFTR gene reliably detects more than one third of non-?F508 mutations in German cystic fibrosis patients. Human Genetics, 1992, 88, 283-7. | 1.8 | 13 |
| 428 | The mutational spectrum of single base-pair substitutions in mRNA splice junctions of human genes: Causes and consequences. Human Genetics, 1992, 90, 41-54. | 1.8 | 1,182 |
| 429 | Protein C deficiency and thromboembolism: recurrent mutation at Arg 306 in the protein C gene. Human Genetics, 1992, 88, 586-588. | 1.8 | 8 |
| 430 | Prenatal exclusion of haemophilia a and carrier testing by direct detection of a disease lesion. Prenatal Diagnosis, 1992, 12, 861-866. | 1.1 | 2 |
| 431 | A novel missense mutation in the antithrombin III gene (Ser349â†'Pro) causing recurrent venous thrombosis. Human Genetics, 1992, 88, 707-708. | 1.8 | 5 |
| 432 | A single base-pair deletion in the protein C gene causing recurrent thromboembolism. Thrombosis Research, 1991, 61, 335-340. | 0.8 | 6 |

| # | Article | IF | Citations |
|---|---|---|---|
| 433 | Late-onset homozygous protein C deficiency. Lancet, The, 1991, 338, 575-576. | 6.3 | 32 |
| 434 | Report of the DNA committee and catalogues of cloned and mapped genes, markers formatted for PCR and DNA polymorphisms (Part 1 of 27). Cytogenetic and Genome Research, 1991, 58, 1190-1211. | 0.6 | 78 |
| 435 | The molecular genetics of familial venous thrombosis. Blood Reviews, 1991, 5, 55-70. | 2.8 | 13 |
| 436 | Carrier detection in haemophilia A by direct analysis of factor VIII gene lesions. Human Genetics, 1991, 87, 99-100. | 1.8 | 2 |
| 437 | Gene deletions causing human genetic disease: mechanisms of mutagenesis and the role of the local DNA sequence environment. Human Genetics, 1991, 86, 425-41. | 1.8 | 438 |
| 438 | Mechanisms of insertional mutagenesis in human genes causing genetic disease. Human Genetics, 1991, 87, 409-15. | 1.8 | 119 |
| 439 | A comprehensive list of cloned human DNA sequences--1990 update. Nucleic Acids Research, 1991, 19, 2111-2126. | 6.5 | 4 |
| 440 | The Molecular Genetics of Platelet Membrane Proteins and their Inherited Disorders. Platelets, 1991, 2, 59-67. | 1.1 | 0 |
| 441 | The mutational spectrum of single base-pair substitutions causing human genetic disease: patterns and predictions. Human Genetics, 1990, 85, 55-74. | 1.8 | 358 |
| 442 | Report of the DNA committee and catalogues of cloned and mapped genes and DNA polymorphisms (Part 1 of 14). Cytogenetic and Genome Research, 1990, 55, 457-472. | 0.6 | 50 |
| 443 | MspI RFLP in the human heparin cofactor II (HCF2) gene. Nucleic Acids Research, 1990, 18, 1664-1664. | 6.5 | 3 |
| 444 | A comprehensive list of cloned human DNA sequences. Nucleic Acids Research, 1990, 18, 2413-2547. | 6.5 | 6 |
| 445 | The effect of replication errors on the mismatch analysis of PCR-amplified DNA. Nucleic Acids Research, 1990, 18, 973-978. | 6.5 | 47 |
| 446 | Molecular genetic analysis of a novel form of haemophilia a characterized by the variable expression of factor VIII. Thrombosis Research, 1990, 59, 871-877. | 0.8 | 7 |
| 447 | Protein C London 1: recurrent mutation at Arg 169 (CGGâ€"TGG) in the protein C gene causing thrombosis. Nucleic Acids Research, 1989, 17, 10513-10513. | 6.5 | 18 |
| 448 | Cytosine methylation and the fate of CpG dinucleotides in vertebrate genomes. Human Genetics, 1989, 83, 181-188. | 1.8 | 303 |
| 449 | Diagnosis of genetic disease using recombinant DNA. Second edition. Human Genetics, 1989, 83, 307-334. | 1.8 | 27 |
| 450 | The CpG dinucleotide and human genetic disease. Human Genetics, 1988, 78, 151-155. | 1.8 | 932 |

| # | Article | IF | Citations |
|---|---|---|---|
| 451 | DNA polymorphism and the study of disease associations. Human Genetics, 1988, 78, 299-312. | 1.8 | 98 |
| 452 | Down's syndrome and the molecular biology of chromosome 21. Progress in Neurobiology, 1988, 30, 507-530. | 2.8 | 32 |
| 453 | Precursor-product relationship between vitellogenin and the yolk proteins as derived from the complete sequence of aXenopusvitellogenin gene. Nucleic Acids Research, 1987, 15, 4737-4760. | 6.5 | 123 |
| 454 | Molecular genetic approaches to the analysis of human ophthalmic disease. Eye, 1987, 1, 699-721. | 1.1 | 3 |
| 455 | The distribution of the dinucleotide CpG and cytosine methylation in the vitellogenin gene family. Journal of Molecular Evolution, 1987, 25, 107-115. | 0.8 | 16 |
| 456 | Localization of a human heat-shock HSP 70 gene sequence to chromosome 6 and detection of two other loci by somatic-cell hybrid and restriction fragment length polymorphism analysis. Human Genetics, 1987, 75, 123-128. | 1.8 | 66 |
| 457 | Regional localization and characterization of a DNA segment on the long arm of chromosome 21. Human Genetics, 1987, 75, 129-135. | 1.8 | 8 |
| 458 | Diagnosis of genetic disease using recombinant DNA. Human Genetics, 1986, 73, 1-11. | 1.8 | 71 |
| 459 | Human gene cloning: the storm before the lull?. Nature, 1986, 322, 119-119. | 13.7 | 8 |
| 460 | An estimate of unique DNA sequence heterozygosity in the human genome. Human Genetics, 1985, 69, 201-205. | 1.8 | 298 |
| 461 | DNA methylation and CpG suppression. Cell Differentiation, 1985, 17, 199-205. | 1.3 | 59 |
| 462 | A list of cloned human DNA sequences-Supplement. Human Genetics, 1984, 67, 111-114. | 1.8 | 13 |
| 463 | DNA restriction fragment length polymorphisms and heterozygosity in the human genome. Human Genetics, 1984, 66, 1-16. | 1.8 | 155 |
| 464 | Restriction fragment length polymorphisms at the human parathyroid hormone gene locus. Human Genetics, 1984, 67, 428-431. | 1.8 | 68 |
| 465 | The Pattern of DNA Methylation in the Î´-Crystallin Genes in Transdifferentiating Neural Retina Cultures. Differentiation, 1983, 24, 33-38. | 1.0 | 13 |
| 466 | A list of cloned human DNA sequences. Human Genetics, 1983, 65, 19-26. | 1.8 | 14 |
| 467 | Eukaryotic DNA methylation. Human Genetics, 1983, 64, 315-333. | 1.8 | 193 |
| 468 | Unmethlated domains in vertebrate DNA. Nucleic Acids Research, 1983, 11, 647-658. | 6.5 | 174 |

| # | Article | IF | Citations |
|---|---|---|---|
| 469 | Chicken lens Î´-crystallin gene expression and methylation in several non-lens tissues. Nucleic Acids Research, 1983, 11, 2513-2527. | 6.5 | 36 |