# Andrew Collins

## List of Publications by Year in Descending Order



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|---|---|---|---|
| 181 papers | 8,115 citations | 45 h-index | 85 g-index |
| 191 ext. papers | 9,048 ext. citations | 6.7 avg, IF | 5.48 L-index |

| # | Paper | IF | Citations |
|---|---|---|---|
| 181 | Clinical and Descriptive Study of Orofacial Clefts in Colombia: 2069 Patients From Operation Smile Foundation. *Cleft Palate-Craniofacial Journal*, **2021**, 10556656211000551 | 1.9 | 3 |
| 180 | Essentiality-specific pathogenicity prioritization gene score to improve filtering of disease sequence data. *Briefings in Bioinformatics*, **2021**, 22, 1782-1789 | 13.4 | 0 |
| 179 | Clinical significance of TP53, BIRC3, ATM and MAPK-ERK genes in chronic lymphocytic leukaemia: data from the randomised UK LRF CLL4 trial. *Leukemia*, **2020**, 34, 1760-1774 | 10.7 | 20 |
| 178 | zalpha: an R package for the identification of regions of the genome under selection. *Journal of Open Source Software*, **2020**, 5, 2638 | 5.2 | |
| 177 | Linkage disequilibrium maps for European and African populations constructed from whole genome sequence data. *Scientific Data*, **2019**, 6, 208 | 8.2 | 3 |
| 176 | Gene-dense autosomal chromosomes show evidence for increased selection. *Heredity*, **2019**, 123, 774-783 | 3.6 | 2 |
| 175 | Heterogeneity in the extent of linkage disequilibrium among exonic, intronic, non-coding RNA and intergenic chromosome regions. *European Journal of Human Genetics*, **2019**, 27, 1436-1444 | 5.3 | 2 |
| 174 | Linkage disequilibrium maps to guide contig ordering for genome assembly. *Bioinformatics*, **2019**, 35, 541-545 | 7.2 | 5 |
| 173 | Sequencing era methods for identifying signatures of selection in the genome. *Briefings in Bioinformatics*, **2019**, 20, 1997-2008 | 13.4 | 8 |
| 172 | Clinical significance of DNA methylation in chronic lymphocytic leukemia patients: results from 3 UK clinical trials. *Blood Advances*, **2019**, 3, 2474-2481 | 7.8 | 13 |
| 171 | Chromosome-level assembly of the water buffalo genome surpasses human and goat genomes in sequence contiguity. *Nature Communications*, **2019**, 10, 260 | 17.4 | 75 |
| 170 | Gene-specific metrics to facilitate identification of disease genes for molecular diagnosis in patient genomes: a systematic review. *Briefings in Functional Genomics*, **2019**, 18, 23-29 | 4.9 | 3 |
| 169 | Understanding the disease genome: gene essentiality and the interplay of selection, recombination and mutation. *Briefings in Bioinformatics*, **2019**, 20, 267-273 | 13.4 | 6 |
| 168 | The Challenge of Genome Sequence Assembly. *Open Bioinformatics Journal*, **2018**, 11, 231-239 | 0.8 | 2 |
| 167 | Machine Learning Approaches: Data Integration for Disease Prediction and Prognosis. *Translational Bioinformatics*, **2018**, 137-141 | | 6 |
| 166 | Analytical Approaches for Exome Sequence Data. *Translational Bioinformatics*, **2018**, 121-136 | | |
| 165 | Single-cell exomes in an index case of amp1q21 multiple myeloma reveal more diverse mutanomes than the whole population. *Blood*, **2018**, 132, 232-235 | 2.2 | 1 |

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| 164 | Evaluating phenotype-driven approaches for genetic diagnoses from exomes in a clinical setting. *Scientific Reports*, **2017**, 7, 13509 | 4.9 | 14 |
| 163 | Germline variation in ADAMTSL1 is associated with prognosis following breast cancer treatment in young women. *Nature Communications*, **2017**, 8, 1632 | 17.4 | 13 |
| 162 | Non-coding NOTCH1 mutations in chronic lymphocytic leukemia; their clinical impact in the UK CLL4 trial. *Leukemia*, **2017**, 31, 510-514 | 10.7 | 21 |
| 161 | Deleterious coding variants in multi-case families with non-syndromic cleft lip and/or palate phenotypes. *Scientific Reports*, **2016**, 6, 30457 | 4.9 | 14 |
| 160 | Exome Sequencing in Classic Hairy Cell Leukaemia Reveals Widespread Variation in Acquired Somatic Mutations between Individual Tumours Apart from the Signature BRAF V(600)E Lesion. *PLoS ONE*, **2016**, 11, e0149162 | 3.7 | 11 |
| 159 | Aarskog-Scott syndrome: phenotypic and genetic heterogeneity. *AIMS Genetics*, **2016**, 03, 049-059 | 2.1 | 1 |
| 158 | Single Cell Whole Exome Sequencing in an Index Case of Amp1q21 Multiple Myeloma to Define Intraclonal Variation. *Blood*, **2016**, 128, 5651-5651 | 2.2 | |
| 157 | Longitudinal copy number, whole exome and targeted deep sequencing of @good riskQ IGHV-mutated CLL patients with progressive disease. *Leukemia*, **2016**, 30, 1301-10 | 10.7 | 32 |
| 156 | Commercial chicken breeds exhibit highly divergent patterns of linkage disequilibrium. *Heredity*, **2016**, 117, 375-382 | 3.6 | 10 |
| 155 | Genetics and Prognostication in Splenic Marginal Zone Lymphoma: Revelations from Deep Sequencing. *Clinical Cancer Research*, **2015**, 21, 4174-4183 | 12.9 | 96 |
| 154 | Telomere length predicts progression and overall survival in chronic lymphocytic leukemia: data from the UK LRF CLL4 trial. *Leukemia*, **2015**, 29, 2411-4 | 10.7 | 38 |
| 153 | Exome sequence read depth methods for identifying copy number changes. *Briefings in Bioinformatics*, **2015**, 16, 380-92 | 13.4 | 55 |
| 152 | The genomic and functional characteristics of disease genes. *Briefings in Bioinformatics*, **2015**, 16, 16-23 | 13.4 | 7 |
| 151 | Quantifying the cumulative effect of low-penetrance genetic variants on breast cancer risk. *Molecular Genetics & Genomic Medicine*, **2015**, 3, 182-8 | 2.3 | 1 |
| 150 | Whole genome sequences are required to fully resolve the linkage disequilibrium structure of human populations. *BMC Genomics*, **2015**, 16, 666 | 4.5 | 12 |
| 149 | Resolving clinical diagnoses for syndromic cleft lip and/or palate phenotypes using whole-exome sequencing. *Clinical Genetics*, **2015**, 88, 441-9 | 4 | 14 |
| 148 | A genome wide meta-analysis study for identification of common variation associated with breast cancer prognosis. *PLoS ONE*, **2014**, 9, e101488 | 3.7 | 24 |
| 147 | ATM mutation rather than BIRC3 deletion and/or mutation predicts reduced survival in 11q-deleted chronic lymphocytic leukemia: data from the UK LRF CLL4 trial. *Haematologica*, **2014**, 99, 736-42 | 6.6 | 59 |

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| 146 | Megalencephaly syndromes: exome pipeline strategies for detecting low-level mosaic mutations. *PLoS ONE*, **2014**, 9, e86940 | 3.7 | 17 |
| 145 | Deep-Sequencing Reveals the Molecular Landscape of Splenic Marginal Zone Lymphoma: Biological and Clinical Implications. *Blood*, **2014**, 124, 76-76 | 2.2 | 1 |
| 144 | Exome sequencing in tracking clonal evolution in multiple myeloma following therapy. *Leukemia*, **2013**, 27, 1188-91 | 10.7 | 17 |
| 143 | Machine learning approaches for the discovery of gene-gene interactions in disease data. *Briefings in Bioinformatics*, **2013**, 14, 251-60 | 13.4 | 60 |
| 142 | A SNP profiling panel for sample tracking in whole-exome sequencing studies. *Genome Medicine*, **2013**, 5, 89 | 14.4 | 45 |
| 141 | The clinical significance of NOTCH1 and SF3B1 mutations in the UK LRF CLL4 trial. *Blood*, **2013**, 121, 468-75 | 11.2 | 167 |
| 140 | Exome-based linkage disequilibrium maps of individual genes: functional clustering and relationship to disease. *Human Genetics*, **2013**, 132, 233-43 | 6.3 | 11 |
| 139 | Identification of inherited genetic variations influencing prognosis in early-onset breast cancer. *Cancer Research*, **2013**, 73, 1883-91 | 10.1 | 36 |
| 138 | Next generation exome sequencing of paediatric inflammatory bowel disease patients identifies rare and novel variants in candidate genes. *Gut*, **2013**, 62, 977-84 | 19.2 | 92 |
| 137 | Support Vector Machine classifier for estrogen receptor positive and negative early-onset breast cancer. *PLoS ONE*, **2013**, 8, e68606 | 3.7 | 13 |
| 136 | Whole exome sequencing identifies novel recurrently mutated genes in patients with splenic marginal zone lymphoma. *PLoS ONE*, **2013**, 8, e83244 | 3.7 | 56 |
| 135 | Variation in complement component C1 inhibitor in age-related macular degeneration. *Immunobiology*, **2012**, 217, 251-5 | 3.4 | 14 |
| 134 | Primer1: Primer Design Web Service for Tetra-Primer ARMS-PCR. *Open Bioinformatics Journal*, **2012**, 6, 55-58 | 0.8 | 62 |
| 133 | The Correlation Between Deletion Architecture, ATM Mutational Status and BIRC3 Disruption in 11q-Deleted CLL. *Blood*, **2012**, 120, 658-658 | 2.2 | 1 |
| 132 | Genome variation: a review of Web resources. *Methods in Molecular Biology*, **2011**, 713, 129-39 | 1.4 | 2 |
| 131 | Genetic variants within chromosome 4q28.3 are not reproducibly associated with age-related macular degeneration (AMD). *Acta Ophthalmologica*, **2011**, 89, e603-4 | 3.7 | |
| 130 | The genetics of breast cancer: risk factors for disease. *The Application of Clinical Genetics*, **2011**, 4, 11-9 | 3.1 | 21 |
| 129 | Genome-wide association of breast cancer: composite likelihood with imputed genotypes. *European Journal of Human Genetics*, **2011**, 19, 194-9 | 5.3 | 5 |

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| 128 | 13q deletion anatomy and disease progression in patients with chronic lymphocytic leukemia. *Leukemia*, **2011**, 25, 489-97 | 10.7 | 93 |
| 127 | Composite likelihood-based meta-analysis of breast cancer association studies. *Journal of Human Genetics*, **2011**, 56, 377-82 | 4.3 | 5 |
| 126 | Support for the involvement of complement factor I in age-related macular degeneration. *European Journal of Human Genetics*, **2010**, 18, 15-6 | 5.3 | 53 |
| 125 | Determination of a gene and environment risk model for age-related macular degeneration. *British Journal of Ophthalmology*, **2010**, 94, 1382-7 | 5.5 | 21 |
| 124 | TFG, a target of chromosome translocations in lymphoma and soft tissue tumors, fuses to GPR128 in healthy individuals. *Haematologica*, **2010**, 95, 20-6 | 6.6 | 45 |
| 123 | The interleukin-1 cluster gene region is associated with multiple sclerosis in an Italian Caucasian population. *European Journal of Neurology*, **2010**, 17, 930-8 | 6 | 17 |
| 122 | Deletion Size Influences Clinical Outcome In Patients with Chronic Lymphocytic Leukemia; 13q Deletion Anatomy, Cooperating Lesions and Cancer Pathogenesis. *Blood*, **2010**, 116, 757-757 | 2.2 | 1 |
| 121 | Mutations in phospholipase C epsilon 1 are not sufficient to cause diffuse mesangial sclerosis. *Kidney International*, **2009**, 75, 415-9 | 9.9 | 32 |
| 120 | Allelic association: linkage disequilibrium structure and gene mapping. *Molecular Biotechnology*, **2009**, 41, 83-9 | 3 | 9 |
| 119 | Approaches to the identification of susceptibility genes. *Parasite Immunology*, **2009**, 31, 225-33 | 2.2 | 8 |
| 118 | Genome-wide association analysis of metabolic traits in a birth cohort from a founder population. *Nature Genetics*, **2009**, 41, 35-46 | 36.3 | 588 |
| 117 | JAK2 haplotype is a major risk factor for the development of myeloproliferative neoplasms. *Nature Genetics*, **2009**, 41, 446-9 | 36.3 | 320 |
| 116 | Extent of genome-wide linkage disequilibrium in Australian Holstein-Friesian cattle based on a high-density SNP panel. *BMC Genomics*, **2008**, 9, 187 | 4.5 | 155 |
| 115 | Linkage disequilibrium in maps of SNPs and other markers. *GeneScreen*, **2008**, 1, 59-61 | | |
| 114 | The influence of genetic variation in 30 selected genes on the clinical characteristics of early onset breast cancer. *Breast Cancer Research*, **2008**, 10, R108 | 8.3 | 42 |
| 113 | Association between the SERPING1 gene and age-related macular degeneration: a two-stage case-control study. *Lancet, The*, **2008**, 372, 1828-34 | 40 | 143 |
| 112 | Individual disease risk and multimetric analysis of Crohn disease. *Proceedings of the National Academy of Sciences of the United States of America*, **2008**, 105, 15843-7 | 11.5 | 3 |
| 111 | A comparison of methods to detect recombination hotspots. *Human Heredity*, **2008**, 66, 157-69 | 1.1 | 7 |

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| 110 | A multimetric approach to analysis of genome-wide association by single markers and composite likelihood. *Proceedings of the National Academy of Sciences of the United States of America*, **2008**, 105, 2592-7 | 11.5 | 6 |
| 109 | CHROMSCAN: genome-wide association using a linkage disequilibrium map. *Journal of Human Genetics*, **2008**, 53, 121-126 | 4.3 | 11 |
| 108 | The genome-wide patterns of variation expose significant substructure in a founder population. *American Journal of Human Genetics*, **2008**, 83, 787-94 | 11 | 116 |
| 107 | Development of V617F JAK2 Associated Myeloproliferative Neoplasms Is a Non-Random Event That Is Strongly Dependent on JAK2 Haplotype. *Blood*, **2008**, 112, 173-173 | 2.2 | |
| 106 | The origin of trisomy 13. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 2242-8 | 2.5 | 26 |
| 105 | Quantitated transcript haplotypes (QTH) of AGTR1, reduced abundance of mRNA haplotypes containing 1166C (rs5186:A>C), and relevance to metabolic syndrome traits. *Human Mutation*, **2007** , 28, 365-73 | 4.7 | 15 |
| 104 | Effects of single SNPs, haplotypes, and whole-genome LD maps on accuracy of association mapping. *Genetic Epidemiology*, **2007**, 31, 179-88 | 2.6 | 15 |
| 103 | The BRCA1 Ashkenazi founder mutations occur on common haplotypes and are not highly correlated with anonymous single nucleotide polymorphisms likely to be used in genome-wide case-control association studies. *BMC Genetics*, **2007**, 8, 68 | 2.6 | 7 |
| 102 | A comparative location database (CompLDB): map integration within and between species. *Mammalian Genome*, **2007**, 18, 287-99 | 3.2 | 14 |
| 101 | Non-disjunction of chromosome 13. *Human Molecular Genetics*, **2007**, 16, 2004-10 | 5.6 | 32 |
| 100 | Fine-scale linkage disequilibrium mapping of age-related macular degeneration in the complement factor H gene region. *British Journal of Ophthalmology*, **2007**, 91, 966-70 | 5.5 | 11 |
| 99 | Exploiting large scale computing to construct high resolution linkage disequilibrium maps of the human genome. *Bioinformatics*, **2007**, 23, 517-9 | 7.2 | 26 |
| 98 | Mapping a gene for rheumatoid arthritis on chromosome 18q21. *BMC Proceedings*, **2007**, 1 Suppl 1, S18 | 2.3 | 5 |
| 97 | Genome scanning by composite likelihood. *American Journal of Human Genetics*, **2007**, 80, 19-28 | 11 | 16 |
| 96 | Linkage disequilibrium and association mapping: an introduction. *Methods in Molecular Biology*, **2007**, 376, 1-15 | 1.4 | 4 |
| 95 | LDMAP: the construction of high-resolution linkage disequilibrium maps of the human genome. *Methods in Molecular Biology*, **2007**, 376, 47-57 | 1.4 | 7 |
| 94 | Extended tracts of homozygosity in outbred human populations. *Human Molecular Genetics*, **2006**, 15, 789-95 | 5.6 | 282 |
| 93 | A first-generation metric linkage disequilibrium map of bovine chromosome 6. *Genetics*, **2006**, 174, 79-85 | 4 | 32 |

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| 92 | Refined association mapping for a quantitative trait: weight in the H19-IGF2-INS-TH region. *Annals of Human Genetics*, **2006**, 70, 848-56 | 2.2 | 17 |
| 91 | Magnitude and distribution of linkage disequilibrium in population isolates and implications for genome-wide association studies. *Nature Genetics*, **2006**, 38, 556-60 | 36.3 | 202 |
| 90 | Cosmopolitan linkage disequilibrium maps. *Human Genomics*, **2005**, 2, 20-7 | 6.8 | 10 |
| 89 | EFFECT OF THE PEROXISOME PROLIFERATORS-ACTIVATED RECEPTOR (PPAR) GAMMA 3 GENE ON BMI IN 1,210 SCHOOL STUDENTS FROM MORELOS, MEXICO **2005**, | | 2 |
| 88 | Complex segregation analysis of nasopharyngeal carcinoma in Guangdong, China: evidence for a multifactorial mode of inheritance (complex segregation analysis of NPC in China). *European Journal of Human Genetics*, **2005**, 13, 248-52 | 5.3 | 43 |
| 87 | Linkage disequilibrium analysis of case-control data: an application to generalized aggressive periodontitis. *Genes and Immunity*, **2005**, 6, 44-52 | 4.4 | 30 |
| 86 | The optimal measure of linkage disequilibrium reduces error in association mapping of affection status. *Human Molecular Genetics*, **2005**, 14, 145-53 | 5.6 | 40 |
| 85 | A map of the human genome in linkage disequilibrium units. *Proceedings of the National Academy of Sciences of the United States of America*, **2005**, 102, 11835-9 | 11.5 | 71 |
| 84 | Polymorphisms in a disintegrin and metalloprotease 33 (ADAM33) predict impaired early-life lung function. *American Journal of Respiratory and Critical Care Medicine*, **2005**, 172, 55-60 | 10.2 | 116 |
| 83 | The linkage disequilibrium maps of three human chromosomes across four populations reflect their demographic history and a common underlying recombination pattern. *Genome Research*, **2005**, 15, 454-62 | 9.7 | 87 |
| 82 | MaGIC: a program to generate targeted marker sets for genome-wide association studies. *BioTechniques*, **2004**, 37, 996-9 | 2.5 | 8 |
| 81 | The impact of SNP density on fine-scale patterns of linkage disequilibrium. *Human Molecular Genetics*, **2004**, 13, 577-88 | 5.6 | 171 |
| 80 | Mapping genes for common diseases: the case for genetic (LD) maps. *Human Heredity*, **2004**, 58, 2-9 | 1.1 | 29 |
| 79 | Impact of population structure, effective bottleneck time, and allele frequency on linkage disequilibrium maps. *Proceedings of the National Academy of Sciences of the United States of America*, **2004**, 101, 18075-80 | 11.5 | 33 |
| 78 | Does haplotype diversity predict power for association mapping of disease susceptibility?. *Human Genetics*, **2004**, 115, 157-64 | 6.3 | 43 |
| 77 | Positional cloning by linkage disequilibrium. *American Journal of Human Genetics*, **2004**, 74, 846-55 | 11 | 50 |
| 76 | Linkage disequilibrium in human populations. *Proceedings of the National Academy of Sciences of the United States of America*, **2003**, 100, 6069-74 | 11.5 | 52 |
| 75 | Haplotypic analysis of the MMP-9 gene in relation to coronary artery disease. *Journal of Molecular Medicine*, **2003**, 81, 321-6 | 5.5 | 83 |

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| 74 | CpG islands in human X-inactivation. *Annals of Human Genetics*, **2003**, 67, 242-9 | 2.2 | 34 |
| 73 | A metric linkage disequilibrium map of a human chromosome. *Annals of Human Genetics*, **2003**, 67, 487-94 | 2.2 | 39 |
| 72 | Independent effects of the -219 G>T and epsilon 2/ epsilon 3/ epsilon 4 polymorphisms in the apolipoprotein E gene on coronary artery disease: the Southampton Atherosclerosis Study. *European Journal of Human Genetics*, **2003**, 11, 437-43 | 5.3 | 36 |
| 71 | Influence of TNFalpha and LTalpha single nucleotide polymorphisms on susceptibility to and prognosis in cutaneous malignant melanoma in the British population. *International Journal of Immunogenetics*, **2002**, 29, 17-23 | | 32 |
| 70 | The power and statistical behaviour of allele-sharing statistics when applied to models with two disease loci. *Journal of Genetics*, **2002**, 81, 99-103 | 1.2 | 2 |
| 69 | The distinguishing sequence characteristics of mouse imprinted genes. *Mammalian Genome*, **2002**, 13, 639-45 | 3.2 | 35 |
| 68 | Complex segregation analysis of hypospadias. *Human Genetics*, **2002**, 111, 231-4 | 6.3 | 44 |
| 67 | A novel approach for identifying candidate imprinted genes through sequence analysis of imprinted and control genes. *Human Genetics*, **2002**, 111, 511-20 | 6.3 | 20 |
| 66 | Recombination, interference and sequence: comparison of chromosomes 21 and 22. *Annals of Human Genetics*, **2002**, 66, 75-86 | 2.2 | 9 |
| 65 | A linkage tournament: affection status, parametric analysis, multivariate traits, and enhancements to variance components and relative pairs. *Annals of Human Genetics*, **2002**, 66, 87-98 | 2.2 | 4 |
| 64 | Influence of vascular endothelial growth factor single nucleotide polymorphisms on tumour development in cutaneous malignant melanoma. *Genes and Immunity*, **2002**, 3, 229-32 | 4.4 | 143 |
| 63 | Genetic analysis of multicase families of visceral leishmaniasis in northeastern Brazil: no major role for class II or class III regions of HLA. *Genes and Immunity*, **2002**, 3, 350-8 | 4.4 | 25 |
| 62 | Properties of linkage disequilibrium (LD) maps. *Proceedings of the National Academy of Sciences of the United States of America*, **2002**, 99, 17004-7 | 11.5 | 71 |
| 61 | The first linkage disequilibrium (LD) maps: delineation of hot and cold blocks by diplotype analysis. *Proceedings of the National Academy of Sciences of the United States of America*, **2002**, 99, 2228-33 | 11.5 | 155 |
| 60 | PCR designer for restriction analysis of various types of sequence mutation. *Bioinformatics*, **2002**, 18, 1688-9 | 7.2 | 7 |
| 59 | Mapping quantitative effects of oligogenes by allelic association. *Annals of Human Genetics*, **2002**, 66, 211-21 | 2.2 | 3 |
| 58 | Allelic association with SNPs: metrics, populations, and the linkage disequilibrium map. *Human Mutation*, **2001**, 17, 255-62 | 4.7 | 28 |
| 57 | Association and linkage of leprosy phenotypes with HLA class II and tumour necrosis factor genes. *Genes and Immunity*, **2001**, 2, 196-204 | 4.4 | 79 |

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| 56 | Maternal sex chromosome non-disjunction: evidence for X chromosome-specific risk factors. *Human Molecular Genetics*, **2001**, 10, 243-50 | 5.6 | 50 |
| 55 | Genetic epidemiology of glioma. *British Journal of Cancer*, **2001**, 84, 429-34 | 8.7 | 57 |
| 54 | The optimal measure of allelic association. *Proceedings of the National Academy of Sciences of the United States of America*, **2001**, 98, 5217-21 | 11.5 | 99 |
| 53 | A sequence-based integrated map of chromosome 22. *Genome Research*, **2001**, 11, 1290-5 | 9.7 | 14 |
| 52 | Allelic association and disease mapping. *Briefings in Bioinformatics*, **2001**, 2, 375-87 | 13.4 | 6 |
| 51 | An efficient procedure for genotyping single nucleotide polymorphisms. *Nucleic Acids Research*, **2001**, 29, E88-8 | 20.1 | 684 |
| 50 | Combination of linkage evidence in complex inheritance. *Human Heredity*, **2001**, 52, 132-5 | 1.1 | 3 |
| 49 | A tournament of linkage tests in complex inheritance. *Human Heredity*, **2001**, 52, 140-8 | 1.1 | 9 |
| 48 | Combined segregation and linkage analysis of 59 Hodgkin disease families indicates the role of HLA determinants. *European Journal of Human Genetics*, **2000**, 8, 460-3 | 5.3 | 6 |
| 47 | A reinvestigation of non-disjunction resulting in 47, XXY males of paternal origin. *European Journal of Human Genetics*, **2000**, 8, 805-8 | 5.3 | 44 |
| 46 | Combined segregation and linkage analysis of inflammatory bowel disease in the IBD1 region using severity to characterise Crohn disease and ulcerative colitis. On behalf of the GISC. *European Journal of Human Genetics*, **2000**, 8, 846-52 | 5.3 | 16 |
| 45 | Mapping in the sequencing era. *Human Heredity*, **2000**, 50, 76-84 | 1.1 | 7 |
| 44 | A novel X-linked dominant condition: X-linked congenital isolated ptosis. *American Journal of Human Genetics*, **2000**, 66, 1455-60 | 11 | 40 |
| 43 | Allelic association between marker loci. *Proceedings of the National Academy of Sciences of the United States of America*, **1999**, 96, 1621-6 | 11.5 | 59 |
| 42 | Genetic epidemiology of single-nucleotide polymorphisms. *Proceedings of the National Academy of Sciences of the United States of America*, **1999**, 96, 15173-7 | 11.5 | 231 |
| 41 | Combined segregation and linkage analysis of nonsyndromic orofacial cleft in two candidate regions. *Annals of Human Genetics*, **1999**, 63, 17-25 | 2.2 | 16 |
| 40 | An evaluation of affected-sib-pair methods and transmission/disequilibrium tests for detecting genes underlying a complex trait. *Genetic Epidemiology*, **1999**, 17 Suppl 1, S727-30 | 2.6 | |
| 39 | The impact of redefining affection status for alcoholism on affected-sib-pair analysis. *Genetic Epidemiology*, **1999**, 17 Suppl 1, S151-6 | 2.6 | 2 |

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| 38 | Linkage of asthma to markers on chromosome 12 in a sample of 240 families using quantitative phenotype scores. *Genomics*, **1998**, 53, 251-9 | 4.3 | 72 |
| 37 | Non-disjunction of chromosome 18. *Human Molecular Genetics*, **1998**, 7, 661-9 | 5.6 | 78 |
| 36 | Tests and estimates of allelic association in complex inheritance. *Proceedings of the National Academy of Sciences of the United States of America*, **1998**, 95, 11389-93 | 11.5 | 214 |
| 35 | Allelic association under map error and recombinational heterogeneity: a tale of two sites. *Proceedings of the National Academy of Sciences of the United States of America*, **1998**, 95, 11366-70 | 11.5 | 31 |
| 34 | Limb girdle muscular dystrophy type 2A (CAPN3): mapping using allelic association. *Human Heredity* , **1998**, 48, 333-7 | 1.1 | 6 |
| 33 | Mapping a disease locus by allelic association. *Proceedings of the National Academy of Sciences of the United States of America*, **1998**, 95, 1741-5 | 11.5 | 144 |
| 32 | Evidence that genetic susceptibility to Mycobacterium tuberculosis in a Brazilian population is under oligogenic control: linkage study of the candidate genes NRAMP1 and TNFA. *Tubercle and Lung Disease*, **1997**, 78, 35-45 | | 105 |
| 31 | The future of gene mapping. *Genetic Analysis, Techniques and Applications*, **1997**, 14, 25-7 | | 1 |
| 30 | A two-locus model for hereditary non-polyposis colorectal cancer in Modena, Italy. *Annals of Human Genetics*, **1997**, 61, 109-19 | 2.2 | 1 |
| 29 | Linkage map integration. *Genomics*, **1996**, 36, 157-62 | 4.3 | 39 |
| 28 | A metric map of humans: 23,500 loci in 850 bands. *Proceedings of the National Academy of Sciences of the United States of America*, **1996**, 93, 14771-5 | 11.5 | 233 |
| 27 | Studies on locus content mapping. *Proceedings of the National Academy of Sciences of the United States of America*, **1996**, 93, 11814-8 | 11.5 | 12 |
| 26 | Trials of the beta model for complex inheritance. *Proceedings of the National Academy of Sciences of the United States of America*, **1996**, 93, 9177-81 | 11.5 | 23 |
| 25 | Complex segregation analysis in a sample of consecutive newborns with cleft lip with or without cleft palate in Italy. *Human Heredity*, **1995**, 45, 157-64 | 1.1 | 24 |
| 24 | Nonparametric tests for linkage with dependent sib pairs. *Human Heredity*, **1995**, 45, 311-8 | 1.1 | 19 |
| 23 | Evolutionary dynamics of the FMR1 locus. *Annals of Human Genetics*, **1995**, 59, 283-9 | 2.2 | 11 |
| 22 | Integration of gene maps: updating chromosome 1. *Annals of Human Genetics*, **1995**, 59, 291-305 | 2.2 | 8 |
| 21 | An integrated map of chromosome 9. *Annals of Human Genetics*, **1995**, 59, 393-402 | 2.2 | 10 |

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| 20 | Exclusion from proximal 11q of a common gene with megaphenic effect on atopy. *Annals of Human Genetics*, **1995**, 59, 403-11 | 2.2 | 22 |
| 19 | Integrated genetic map of human chromosome 2. *Annals of Human Genetics*, **1995**, 59, 413-34 | 2.2 | 5 |
| 18 | Segregation analysis of colorectal cancer in Northern Ireland. *Human Heredity*, **1995**, 45, 41-8 | 1.1 | 9 |
| 17 | An n-allele model for progressive amplification in the FMR1 locus. *Proceedings of the National Academy of Sciences of the United States of America*, **1995**, 92, 4833-7 | 11.5 | 32 |
| 16 | Statistical and genetic aspects of quality control for DNA identification. *Electrophoresis*, **1995**, 16, 1670-7 | 3.6 | 10 |
| 15 | Genetic epidemiology of early onset breast cancer. *Journal of Medical Genetics*, **1994**, 31, 944-9 | 5.8 | 17 |
| 14 | Coding of pointers in the segregation analysis program POINTER. *Genetic Epidemiology*, **1994**, 11, 385-7 | 2.6 | 3 |
| 13 | CEPH consortium Map of chromosome 9. *Genomics*, **1994**, 19, 203-14 | 4.3 | 19 |
| 12 | Integration of gene maps: chromosome X. *Genomics*, **1994**, 22, 590-604 | 4.3 | 27 |
| 11 | Genetic epidemiology of hereditary non-polyposis colorectal cancer syndromes in Modena, Italy: results of a complex segregation analysis. *Annals of Human Genetics*, **1994**, 58, 275-95 | 2.2 | 20 |
| 10 | Likelihood ratios for DNA identification. *Proceedings of the National Academy of Sciences of the United States of America*, **1994**, 91, 6007-11 | 11.5 | 20 |
| 9 | Kinship bioassay on hypervariable loci in blacks and Caucasians. *Proceedings of the National Academy of Sciences of the United States of America*, **1993**, 90, 1892-6 | 11.5 | 39 |
| 8 | Integration of gene maps: chromosome 21. *Proceedings of the National Academy of Sciences of the United States of America*, **1993**, 90, 7210-4 | 11.5 | 24 |
| 7 | Integration of gene maps: chromosome 1. *Proceedings of the National Academy of Sciences of the United States of America*, **1992**, 89, 4598-602 | 11.5 | 35 |
| 6 | Algorithms for a location database. *Annals of Human Genetics*, **1992**, 56, 223-32 | 2.2 | 52 |
| 5 | Error filtration, interference, and the human linkage map. *Proceedings of the National Academy of Sciences of the United States of America*, **1991**, 88, 6501-5 | 11.5 | 60 |
| 4 | Significance of maximal lods. *Annals of Human Genetics*, **1991**, 55, 39-41 | 2.2 | 6 |
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