Edward J Hollox

List of Publications by Year
in descending order

77 papers	3,558 citations	185998 28 h-index	138251 58 g-index
92 all docs	92 docs citations	92 times ranked	4741 citing authors

#	Article	IF	Citations
1	Psoriasis is associated with increased Î²-defensin genomic copy number. Nature Genetics, 2008, 40, 23-25.	9.4	587
2	Extensive Normal Copy Number Variation of a Î²-Defensin Antimicrobial-Gene Cluster. American Journal of Human Genetics, 2003, 73, 591-600.	2.6	315
3	Lactase Haplotype Diversity in the Old World. American Journal of Human Genetics, 2001, 68, 160-172.	2.6	180
4	Copy number polymorphism and expression level variation of the human Î±-defensin genes DEFA1 and DEFA3. Human Molecular Genetics, 2005, 14, 2045-2052.	1.4	174
5	Î²-Defensin-2 Protein Is a Serum Biomarker for Disease Activity in Psoriasis and Reaches Biologically Relevant Concentrations in Lesional Skin. PLoS ONE, 2009, 4, e4725.	1.1	151
6	Accurate, high-throughput typing of copy number variation using paralogue ratios from dispersed repeats. Nucleic Acids Research, 2007, 35, e19-e19.	6.5	128
7	A Common Mutation in the Defensin DEFB126 Causes Impaired Sperm Function and Subfertility. Science Translational Medicine, 2011, 3, 92ra65.	5.8	127
8	The genetically programmed down-regulation of lactase in children. Gastroenterology, 1998, 114, 1230-1236.	0.6	121
9	The Causal Element for the Lactase Persistence/ non-persistence Polymorphism is Located in a 1 Mb Region of Linkage Disequilibrium in Europeans. Annals of Human Genetics, 2003, 67, 298-311.	0.3	109
10	Defensins and the dynamic genome: What we can learn from structural variation at human chromosome band 8p23.1. Genome Research, 2008, 18, 1686-1697.	2.4	79
11	FcÎ³ receptors: genetic variation, function, and disease. Immunological Reviews, 2015, 268, 6-24.	2.8	78
12	Haptoglobin (HP) and Haptoglobin-related protein (HPR) copy number variation, natural selection, and trypanosomiasis. Human Genetics, 2014, 133, 69-83.	1.8	72
13	Clonal architecture in mesothelioma is prognostic and shapes the tumour microenvironment. Nature Communications, 2021, 12, 1751.	5.8	66
14	A worldwide analysis of beta-defensin copy number variation suggests recent selection of a high-expressing DEFB103 gene copy in East Asia. Human Mutation, 2011, 32, 743-750.	1.1	65
15	Human gene copy number variation and infectious disease. Human Genetics, 2014, 133, 1217-1233.	1.8	63
16	Copy number variation of beta-defensins and relevance to disease. Cytogenetic and Genome Research, 2008, 123, 148-155.	0.6	60
17	An integrated approach for measuring copy number variation at theFCGR3(CD16) locus. Human Mutation, 2009, 30, 477-484.	1.1	60
18	Directional and balancing selection in human beta-defensins. BMC Evolutionary Biology, 2008, 8, 113.	3.2	58

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19	Allelic recombination between distinct genomic locations generates copy number diversity in human Î²-defensins. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 853-858.	3.3	55
20	Lactase haplotype frequencies in Caucasians: association with the lactase persistence/non-persistence polymorphism. Annals of Human Genetics, 1998, 62, 215-223.	0.3	51
21	High throughput screening of human subtelomeric DNA for copy number changes using multiplex amplifiable probe hybridisation (MAPH). Journal of Medical Genetics, 2002, 39, 790-795.	1.5	46
22	Duplications and copy number variants of 8p23.1 are cytogenetically indistinguishable but distinct at the molecular level. European Journal of Human Genetics, 2005, 13, 1131-1136.	1.4	46
23	Determination of Beta-Defensin Genomic Copy Number in Different Populations: A Comparison of Three Methods. PLoS ONE, 2011, 6, e16768.	1.1	39
24	Evolutionary History of Copy-Number-Variable Locus for the Low-Affinity FcÎ³ Receptor: Mutation Rate, Autoimmune Disease, and the Legacy of Helminth Infection. American Journal of Human Genetics, 2012, 90, 973-985.	2.6	38
25	Copy-number variation of the neuronal glucose transporter gene SLC2A3 and age of onset in Huntington's disease. Human Molecular Genetics, 2014, 23, 3129-3137.	1.4	38
26	A 4q35.2 subtelomeric deletion identified in a screen of patients with co-morbid psychiatric illness and mental retardation. BMC Medical Genetics, 2004, 5, 21.	2.1	35
27	Evolution of the rapidly mutating human salivary agglutinin gene (DMBT1) and population subsistence strategy. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 5105-5110.	3.3	35
28	Â-defensin Genomic Copy Number Is Associated With HIV Load and Immune Reconstitution in Sub-Saharan Africans. Journal of Infectious Diseases, 2012, 206, 1012-1019.	1.9	33
29	Genome structural variation in human evolution. Trends in Genetics, 2022, 38, 45-58.	2.9	32
30	Beta-defensin genomic copy number is not a modifier locus for cystic fibrosis. Journal of Negative Results in BioMedicine, 2005, 4, 9.	1.4	29
31	Whole Exome Re-Sequencing Implicates CCDC38 and Cilia Structure and Function in Resistance to Smoking Related Airflow Obstruction. PLoS Genetics, 2014, 10, e1004314.	1.5	29
32	Constitutional trisomy 8 and BehÃ§et syndrome. American Journal of Medical Genetics, Part A, 2009, 149A, 982-986.	0.7	26
33	Complement receptor 1 gene (CR1) intragenic duplication and risk of Alzheimerâ€™s disease. Human Genetics, 2018, 137, 305-314.	1.8	25
34	DNA copy number analysis by MAPH: molecular diagnostic applications. Expert Review of Molecular Diagnostics, 2002, 2, 370-378.	1.5	23
35	Evolutionary Genetics: Genetics of lactase persistence â€“ fresh lessons in the history of milk drinking. European Journal of Human Genetics, 2005, 13, 267-269.	1.4	23
36	Assessment of complement C4 gene copy number using the paralog ratio test. Human Mutation, 2010, 31, 866-874.	1.1	23

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37	Î²-Defensins and Crohn's Disease: Confusion From Counting Copies. American Journal of Gastroenterology, 2010, 105, 360-362.	0.2	23
38	Common polymorphism in a highly variable region upstream of the human lactase gene affects DNA-protein interactions. European Journal of Human Genetics, 1999, 7, 791-800.	1.4	22
39	Understanding the Genomic Structure of Copy-Number Variation of the Low-Affinity FcÎ³ Receptor Region Allows Confirmation of the Association ofFCGR3BDeletion with Rheumatoid Arthritis. Human Mutation, 2017, 38, 390-399.	1.1	21
40	The Malaria-Protective Human Glycophorin Structural Variant DUP4 Shows Somatic Mosaicism and Association with Hemoglobin Levels. American Journal of Human Genetics, 2018, 103, 769-776.	2.6	21
41	CCL3L1 copy number, HIV load, and immune reconstitution in sub-Saharan Africans. BMC Infectious Diseases, 2013, 13, 536.	1.3	20
42	Copy Number Variation of the Beta Defensin Gene Cluster on Chromosome 8p Influences the Bacterial Microbiota within the Nasopharynx of Otitis-Prone Children. PLoS ONE, 2014, 9, e98269.	1.1	19
43	Evidence of Convergent Evolution in Humans and Macaques Supports an Adaptive Role for Copy Number Variation of the Î²-Defensin-2 Gene. Genome Biology and Evolution, 2014, 6, 3025-3038.	1.1	19
44	Human beta defensin (HBD) gene copy number affects HBD2 protein levels: impact on cervical bactericidal immunity in pregnancy. European Journal of Human Genetics, 2018, 26, 434-439.	1.4	19
45	Copy Number Variation of Fc Gamma Receptor Genes in HIV-Infected and HIV-Tuberculosis Co-Infected Individuals in Sub-Saharan Africa. PLoS ONE, 2013, 8, e78165.	1.1	18
46	Evaluation of High-Throughput Genomic Assays for the Fc Gamma Receptor Locus. PLoS ONE, 2015, 10, e0142379.	1.1	17
47	Evolutionary genetics of the human Rh blood group system. Human Genetics, 2012, 131, 1205-1216.	1.8	16
48	The Challenges of Studying Complex and Dynamic Regions of the Human Genome. Methods in Molecular Biology, 2012, 838, 187-207.	0.4	13
49	Analysis of Copy Number Variation Using the Paralogue Ratio Test (PRT). Methods in Molecular Biology, 2017, 1492, 127-146.	0.4	13
50	Human intelectin-1 (ITLN1) genetic variation and intestinal expression. Scientific Reports, 2021, 11, 12889.	1.6	13
51	Immunocytochemical detection of <scp>ERG</scp> expression in exfoliated urinary cells identifies with high specificity patients with prostate cancer. BJU International, 2016, 117, 686-696.	1.3	12
52	A Comparison of Assays for Accurate Copy Number Measurement of the Low-Affinity Fc Gamma Receptor Genes FCGR3A and FCGR3B. PLoS ONE, 2015, 10, e0116791.	1.1	12
53	Copy Number Variation of the Beta-Defensin Genes in Europeans: No Supporting Evidence for Association with Lung Function, Chronic Obstructive Pulmonary Disease or Asthma. PLoS ONE, 2014, 9, e84192.	1.1	11
54	Expansion of a 12-kb VNTR containing the REXO1L1 gene cluster underlies the microscopically visible euchromatic variant of 8q21.2. European Journal of Human Genetics, 2014, 22, 458-463.	1.4	10

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55	Copy number variation of scavenger-receptor cysteine-rich domains within DMBT1 and Crohnâ€™s disease. European Journal of Human Genetics, 2016, 24, 1294-1300.	1.4	10
56	Human CCL3L1 copy number variation, gene expression, and the role of the CCL3L1-CCR5 axis in lung function. Wellcome Open Research, 2018, 3, 13.	0.9	10
57	Human intelectinâ€2 (ITLN2) is selectively expressed by secretory Paneth cells. FASEB Journal, 2022, 36, e22200.	0.2	10
58	Detection of Copy Number Changes in DNA from Formalin Fixed Paraffin Embedded Tissues Using Paralogue Ratio Tests. Analytical Chemistry, 2011, 83, 3484-3492.	3.2	9
59	Cohort Profile: Extended Cohort for E-health, Environment and DNA (EXCEED). International Journal of Epidemiology, 2019, 48, 678-679j.	0.9	9
60	Automated design of paralogue ratio test assays for the accurate and rapid typing of copy number variation. Bioinformatics, 2013, 29, 1997-2003.	1.8	8
61	Population genetics of immune-related multilocus copy number variation in Native Americans. Journal of the Royal Society Interface, 2017, 14, 20170057.	1.5	8
62	Structural variation of the malaria-associated human glycophorin A-B-E region. BMC Genomics, 2020, 21, 446.	1.2	7
63	No evidence for DNA copy number change associated with the DUP25 cytogenetic phenotype. European Journal of Human Genetics, 2003, 11, 911-912.	1.4	6
64	Extensive variation in the intelectin gene family in laboratory and wild mouse strains. Scientific Reports, 2021, 11, 15548.	1.6	6
65	Exome-wide analysis of copy number variation shows association of the human leukocyte antigen region with asthma in UK Biobank. BMC Medical Genomics, 2022, 15, .	0.7	6
66	Evolution and Diversity of Defensins in Vertebrates. , 2017, , 27-50.		5
67	Deleted in malignant brain tumor 1 genetic variation confers urinary tract infection risk in children and mice. Clinical and Translational Medicine, 2021, 11, e477.	1.7	5
68	Recurrent mutation at the classical haptoglobin structural polymorphism. Nature Genetics, 2016, 48, 347-348.	9.4	4
69	No Evidence for Association of Î²â€Defensin Genomic Copy Number with HIV Susceptibility, HIV Load during Clinical Latency, or Progression to AIDS. Annals of Human Genetics, 2017, 81, 27-34.	0.3	4
70	Balancing selection at the human salivary agglutinin gene (DMBT1) driven by host-microbe interactions. IScience, 2022, 25, 104189.	1.9	4
71	Determining multiallelic complex copy number and sequence variation from high coverage exome sequencing data. BMC Genomics, 2015, 16, 891.	1.2	3
72	Analysis of copy number variation at DMBT1 and age-related macular degeneration. BMC Medical Genetics, 2016, 17, 44.	2.1	3

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73	Variation of rs3754689 at lactase gene and inhibitors in admixed Brazilian patients with hemophilia A. Haematologica, 2019, 104, e527-e529.	1.7	2
74	Î²-Defensin Genomic Copy Number Does Not Influence the Age of Onset in Huntington's Disease. Journal of Huntington's Disease, 2013, 2, 107-124.	0.9	1
75	Copy number variation of the REXO1L1 gene cluster; euchromatic deletion variant or susceptibility factor?. European Journal of Human Genetics, 2017, 25, 8-9.	1.4	1
76	Genotyping complex structural variation at the malariaâ€associated human glycophorin locus using a PCRâ€based strategy. Annals of Human Genetics, 2021, 85, 7-17.	0.3	1
77	Human CCL3L1 copy number variation, gene expression, and the role of the CCL3L1-CCR5 axis in lung function. Wellcome Open Research, 0, 3, 13.	0.9	1