Edward J Hollox

List of Publications by Year
in descending order

77 papers	3,558 citations	186265 28 h-index	138484 58 g-index
92 all docs	92 docs citations	92 times ranked	4741 citing authors

#	Article	IF	Citations
1	Genome structural variation in human evolution. Trends in Genetics, 2022, 38, 45-58.	6.7	32
2	Human intelectinâ€2 (ITLN2) is selectively expressed by secretory Paneth cells. FASEB Journal, 2022, 36, e22200.	0.5	10
3	Balancing selection at the human salivary agglutinin gene (DMBT1) driven by host-microbe interactions. IScience, 2022, 25, 104189.	4.1	4
4	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, .	1.5	6
5	Genotyping complex structural variation at the malariaâ€associated human glycophorin locus using a PCRâ€based strategy. Annals of Human Genetics, 2021, 85, 7-17.	0.8	1
6	Clonal architecture in mesothelioma is prognostic and shapes the tumour microenvironment. Nature Communications, 2021, 12, 1751.	12.8	66
7	Human intelectin-1 (ITLN1) genetic variation and intestinal expression. Scientific Reports, 2021, 11, 12889.	3.3	13
8	Deleted in malignant brain tumor 1 genetic variation confers urinary tract infection risk in children and mice. Clinical and Translational Medicine, 2021, 11, e477.	4.0	5
9	Extensive variation in the intelectin gene family in laboratory and wild mouse strains. Scientific Reports, 2021, 11, 15548.	3.3	6
10	Structural variation of the malaria-associated human glycophorin A-B-E region. BMC Genomics, 2020, 21, 446.	2.8	7
11	Cohort Profile: Extended Cohort for E-health, Environment and DNA (EXCEED). International Journal of Epidemiology, 2019, 48, 678-679j.	1.9	9
12	Variation of rs3754689 at lactase gene and inhibitors in admixed Brazilian patients with hemophilia A. Haematologica, 2019, 104, e527-e529.	3.5	2
13	Complement receptor 1 gene (CR1) intragenic duplication and risk of Alzheimerâ€™s disease. Human Genetics, 2018, 137, 305-314.	3.8	25
14	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.	2.8	19
15	Human CCL3L1 copy number variation, gene expression, and the role of the CCL3L1-CCR5 axis in lung function. Wellcome Open Research, 2018, 3, 13.	1.8	10
16	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.	6.2	21
17	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.	0.8	4
18	Population genetics of immune-related multilocus copy number variation in Native Americans. Journal of the Royal Society Interface, 2017, 14, 20170057.	3.4	8

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19	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.	2.5	21
20	Evolution and Diversity of Defensins in Vertebrates. , 2017, , 27-50.		5
21	Analysis of Copy Number Variation Using the Paralogue Ratio Test (PRT). Methods in Molecular Biology, 2017, 1492, 127-146.	0.9	13
22	Copy number variation of the REXO1L1 gene cluster; euchromatic deletion variant or susceptibility factor?. European Journal of Human Genetics, 2017, 25, 8-9.	2.8	1
23	Analysis of copy number variation at DMBT1 and age-related macular degeneration. BMC Medical Genetics, 2016, 17, 44.	2.1	3
24	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.	2.5	12
25	Recurrent mutation at the classical haptoglobin structural polymorphism. Nature Genetics, 2016, 48, 347-348.	21.4	4
26	Copy number variation of scavenger-receptor cysteine-rich domains within DMBT1 and Crohnâ€™s disease. European Journal of Human Genetics, 2016, 24, 1294-1300.	2.8	10
27	FcÎ³ receptors: genetic variation, function, and disease. Immunological Reviews, 2015, 268, 6-24.	6.0	78
28	Determining multiallelic complex copy number and sequence variation from high coverage exome sequencing data. BMC Genomics, 2015, 16, 891.	2.8	3
29	Evaluation of High-Throughput Genomic Assays for the Fc Gamma Receptor Locus. PLoS ONE, 2015, 10, e0142379.	2.5	17
30	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.	7.1	35
31	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.	2.5	12
32	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.	2.5	19
33	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.	2.5	19
34	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.	2.8	10
35	Whole Exome Re-Sequencing Implicates CCDC38 and Cilia Structure and Function in Resistance to Smoking Related Airflow Obstruction. PLoS Genetics, 2014, 10, e1004314.	3.5	29
36	Haptoglobin (HP) and Haptoglobin-related protein (HPR) copy number variation, natural selection, and trypanosomiasis. Human Genetics, 2014, 133, 69-83.	3.8	72

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37	Human gene copy number variation and infectious disease. Human Genetics, 2014, 133, 1217-1233.	3.8	63
38	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.	2.9	38
39	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.	2.5	11
40	CCL3L1 copy number, HIV load, and immune reconstitution in sub-Saharan Africans. BMC Infectious Diseases, 2013, 13, 536.	2.9	20
41	Automated design of paralogue ratio test assays for the accurate and rapid typing of copy number variation. Bioinformatics, 2013, 29, 1997-2003.	4.1	8
42	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.	1.9	1
43	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.	2.5	18
44	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.	4.0	33
45	The Challenges of Studying Complex and Dynamic Regions of the Human Genome. Methods in Molecular Biology, 2012, 838, 187-207.	0.9	13
46	Evolutionary genetics of the human Rh blood group system. Human Genetics, 2012, 131, 1205-1216.	3.8	16
47	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.	6.2	38
48	Detection of Copy Number Changes in DNA from Formalin Fixed Paraffin Embedded Tissues Using Paralogue Ratio Tests. Analytical Chemistry, 2011, 83, 3484-3492.	6.5	9
49	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.	2.5	65
50	A Common Mutation in the Defensin DEFB126 Causes Impaired Sperm Function and Subfertility. Science Translational Medicine, 2011, 3, 92ra65.	12.4	127
51	Determination of Beta-Defensin Genomic Copy Number in Different Populations: A Comparison of Three Methods. PLoS ONE, 2011, 6, e16768.	2.5	39
52	Assessment of complement C4 gene copy number using the paralog ratio test. Human Mutation, 2010, 31, 866-874.	2.5	23
53	Î²-Defensins and Crohn's Disease: Confusion From Counting Copies. American Journal of Gastroenterology, 2010, 105, 360-362.	0.4	23
54	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.	7.1	55

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55	Constitutional trisomy 8 and BehÃ§et syndrome. American Journal of Medical Genetics, Part A, 2009, 149A, 982-986.	1.2	26
56	An integrated approach for measuring copy number variation at theFCGR3(CD16) locus. Human Mutation, 2009, 30, 477-484.	2.5	60
57	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.	2.5	151
58	Psoriasis is associated with increased Î²-defensin genomic copy number. Nature Genetics, 2008, 40, 23-25.	21.4	587
59	Directional and balancing selection in human beta-defensins. BMC Evolutionary Biology, 2008, 8, 113.	3.2	58
60	Copy number variation of beta-defensins and relevance to disease. Cytogenetic and Genome Research, 2008, 123, 148-155.	1.1	60
61	Defensins and the dynamic genome: What we can learn from structural variation at human chromosome band 8p23.1. Genome Research, 2008, 18, 1686-1697.	5.5	79
62	Accurate, high-throughput typing of copy number variation using paralogue ratios from dispersed repeats. Nucleic Acids Research, 2007, 35, e19-e19.	14.5	128
63	Evolutionary Genetics: Genetics of lactase persistence â€“ fresh lessons in the history of milk drinking. European Journal of Human Genetics, 2005, 13, 267-269.	2.8	23
64	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.	2.8	46
65	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
66	Copy number polymorphism and expression level variation of the human Î±-defensin genes DEFA1 and DEFA3. Human Molecular Genetics, 2005, 14, 2045-2052.	2.9	174
67	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
68	The Causal Element for the Lactase Persistence/ nonâ€persistence Polymorphism is Located in a 1 Mb Region of Linkage Disequilibrium in Europeans. Annals of Human Genetics, 2003, 67, 298-311.	0.8	109
69	No evidence for DNA copy number change associated with the DUP25 cytogenetic phenotype. European Journal of Human Genetics, 2003, 11, 911-912.	2.8	6
70	Extensive Normal Copy Number Variation of a Î²-Defensin Antimicrobial-Gene Cluster. American Journal of Human Genetics, 2003, 73, 591-600.	6.2	315
71	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.	3.2	46
72	DNA copy number analysis by MAPH: molecular diagnostic applications. Expert Review of Molecular Diagnostics, 2002, 2, 370-378.	3.1	23

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73	Lactase Haplotype Diversity in the Old World. American Journal of Human Genetics, 2001, 68, 160-172.	6.2	180
74	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.	2.8	22
75	Lactase haplotype frequencies in Caucasians: association with the lactase persistence/nonâ€persistence polymorphism. Annals of Human Genetics, 1998, 62, 215-223.	0.8	51
76	The genetically programmed down-regulation of lactase in children. Gastroenterology, 1998, 114, 1230-1236.	1.3	121
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.	1.8	1