

# Edward J Hollox

## List of Publications by Year in descending order

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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	Psoriasis is associated with increased $\hat{\imath}^2$ -defensin genomic copy number. <i>Nature Genetics</i> , 2008, 40, 23-25.	21.4	587
2	Extensive Normal Copy Number Variation of a $\hat{\imath}^2$ -Defensin Antimicrobial-Gene Cluster. <i>American Journal of Human Genetics</i> , 2003, 73, 591-600.	6.2	315
3	Lactase Haplotype Diversity in the Old World. <i>American Journal of Human Genetics</i> , 2001, 68, 160-172.	6.2	180
4	Copy number polymorphism and expression level variation of the human $\hat{\imath}^{\pm}$ -defensin genes DEFA1 and DEFA3. <i>Human Molecular Genetics</i> , 2005, 14, 2045-2052.	2.9	174
5	$\hat{\imath}^2$ -Defensin-2 Protein Is a Serum Biomarker for Disease Activity in Psoriasis and Reaches Biologically Relevant Concentrations in Lesional Skin. <i>PLoS ONE</i> , 2009, 4, e4725.	2.5	151
6	Accurate, high-throughput typing of copy number variation using paralogue ratios from dispersed repeats. <i>Nucleic Acids Research</i> , 2007, 35, e19-e19.	14.5	128
7	A Common Mutation in the Defensin <i>DEFB126</i> Causes Impaired Sperm Function and Subfertility. <i>Science Translational Medicine</i> , 2011, 3, 92ra65.	12.4	127
8	The genetically programmed down-regulation of lactase in children. <i>Gastroenterology</i> , 1998, 114, 1230-1236.	1.3	121
9	The Causal Element for the Lactase Persistence/ non-persistence Polymorphism is Located in a 1 Mb Region of Linkage Disequilibrium in Europeans. <i>Annals of Human Genetics</i> , 2003, 67, 298-311.	0.8	109
10	Defensins and the dynamic genome: What we can learn from structural variation at human chromosome band 8p23.1. <i>Genome Research</i> , 2008, 18, 1686-1697.	5.5	79
11	Fc $\hat{\imath}^3$ receptors: genetic variation, function, and disease. <i>Immunological Reviews</i> , 2015, 268, 6-24.	6.0	78
12	Haptoglobin (HP) and Haptoglobin-related protein (HPR) copy number variation, natural selection, and trypanosomiasis. <i>Human Genetics</i> , 2014, 133, 69-83.	3.8	72
13	Clonal architecture in mesothelioma is prognostic and shapes the tumour microenvironment. <i>Nature Communications</i> , 2021, 12, 1751.	12.8	66
14	A worldwide analysis of beta-defensin copy number variation suggests recent selection of a high-expressing DEFB103 gene copy in East Asia. <i>Human Mutation</i> , 2011, 32, 743-750.	2.5	65
15	Human gene copy number variation and infectious disease. <i>Human Genetics</i> , 2014, 133, 1217-1233.	3.8	63
16	Copy number variation of beta-defensins and relevance to disease. <i>Cytogenetic and Genome Research</i> , 2008, 123, 148-155.	1.1	60
17	An integrated approach for measuring copy number variation at the <i>FCGR3</i> (CD16) locus. <i>Human Mutation</i> , 2009, 30, 477-484.	2.5	60
18	Directional and balancing selection in human beta-defensins. <i>BMC Evolutionary Biology</i> , 2008, 8, 113.	3.2	58

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19	Allelic recombination between distinct genomic locations generates copy number diversity in human $\beta$ -defensins. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2009, 106, 853-858.	7.1	55
20	Lactase haplotype frequencies in Caucasians: association with the lactase persistence/non-persistence polymorphism. <i>Annals of Human Genetics</i> , 1998, 62, 215-223.	0.8	51
21	High throughput screening of human subtelomeric DNA for copy number changes using multiplex amplifiable probe hybridisation (MAPH). <i>Journal of Medical Genetics</i> , 2002, 39, 790-795.	3.2	46
22	Duplications and copy number variants of 8p23.1 are cytogenetically indistinguishable but distinct at the molecular level. <i>European Journal of Human Genetics</i> , 2005, 13, 1131-1136.	2.8	46
23	Determination of Beta-Defensin Genomic Copy Number in Different Populations: A Comparison of Three Methods. <i>PLoS ONE</i> , 2011, 6, e16768.	2.5	39
24	Evolutionary History of Copy-Number-Variable Locus for the Low-Affinity Fc $\gamma$ 3 Receptor: Mutation Rate, Autoimmune Disease, and the Legacy of Helminth Infection. <i>American Journal of Human Genetics</i> , 2012, 90, 973-985.	6.2	38
25	Copy-number variation of the neuronal glucose transporter gene SLC2A3 and age of onset in Huntington's disease. <i>Human Molecular Genetics</i> , 2014, 23, 3129-3137.	2.9	38
26	A 4q35.2 subtelomeric deletion identified in a screen of patients with co-morbid psychiatric illness and mental retardation. <i>BMC Medical Genetics</i> , 2004, 5, 21.	2.1	35
27	Evolution of the rapidly mutating human salivary agglutinin gene ( <i>DMBT1</i> ) and population subsistence strategy. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, 5105-5110.	7.1	35
28	$\beta$ -defensin Genomic Copy Number Is Associated With HIV Load and Immune Reconstitution in Sub-Saharan Africans. <i>Journal of Infectious Diseases</i> , 2012, 206, 1012-1019.	4.0	33
29	Genome structural variation in human evolution. <i>Trends in Genetics</i> , 2022, 38, 45-58.	6.7	32
30	Beta-defensin genomic copy number is not a modifier locus for cystic fibrosis. <i>Journal of Negative Results in BioMedicine</i> , 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. <i>PLoS Genetics</i> , 2014, 10, e1004314.	3.5	29
32	Constitutional trisomy 8 and Behçet syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 982-986.	1.2	26
33	Complement receptor 1 gene (CR1) intragenic duplication and risk of Alzheimer's disease. <i>Human Genetics</i> , 2018, 137, 305-314.	3.8	25
34	DNA copy number analysis by MAPH: molecular diagnostic applications. <i>Expert Review of Molecular Diagnostics</i> , 2002, 2, 370-378.	3.1	23
35	Evolutionary Genetics: Genetics of lactase persistence – fresh lessons in the history of milk drinking. <i>European Journal of Human Genetics</i> , 2005, 13, 267-269.	2.8	23
36	Assessment of complement C4 gene copy number using the paralog ratio test. <i>Human Mutation</i> , 2010, 31, 866-874.	2.5	23

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37	Î²-Defensins and Crohn's Disease: Confusion From Counting Copies. <i>American Journal of Gastroenterology</i> , 2010, 105, 360-362.	0.4	23
38	Common polymorphism in a highly variable region upstream of the human lactase gene affects DNA-protein interactions. <i>European Journal of Human Genetics</i> , 1999, 7, 791-800.	2.8	22
39	Understanding the Genomic Structure of Copy-Number Variation of the Low-Affinity FcÎ³3 Receptor Region Allows Confirmation of the Association of FCGR3B Deletion with Rheumatoid Arthritis. <i>Human Mutation</i> , 2017, 38, 390-399.	2.5	21
40	The Malaria-Protective Human Glycophorin Structural Variant DUP4 Shows Somatic Mosaicism and Association with Hemoglobin Levels. <i>American Journal of Human Genetics</i> , 2018, 103, 769-776.	6.2	21
41	CCL3L1 copy number, HIV load, and immune reconstitution in sub-Saharan Africans. <i>BMC Infectious Diseases</i> , 2013, 13, 536.	2.9	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. <i>PLoS ONE</i> , 2014, 9, e98269.	2.5	19
43	Evidence of Convergent Evolution in Humans and Macaques Supports an Adaptive Role for Copy Number Variation of the Î²-Defensin-2 Gene. <i>Genome Biology and Evolution</i> , 2014, 6, 3025-3038.	2.5	19
44	Human beta defensin (HBD) gene copy number affects HBD2 protein levels: impact on cervical bactericidal immunity in pregnancy. <i>European Journal of Human Genetics</i> , 2018, 26, 434-439.	2.8	19
45	Copy Number Variation of Fc Gamma Receptor Genes in HIV-Infected and HIV-Tuberculosis Co-Infected Individuals in Sub-Saharan Africa. <i>PLoS ONE</i> , 2013, 8, e78165.	2.5	18
46	Evaluation of High-Throughput Genomic Assays for the Fc Gamma Receptor Locus. <i>PLoS ONE</i> , 2015, 10, e0142379.	2.5	17
47	Evolutionary genetics of the human Rh blood group system. <i>Human Genetics</i> , 2012, 131, 1205-1216.	3.8	16
48	The Challenges of Studying Complex and Dynamic Regions of the Human Genome. <i>Methods in Molecular Biology</i> , 2012, 838, 187-207.	0.9	13
49	Analysis of Copy Number Variation Using the Parologue Ratio Test (PRT). <i>Methods in Molecular Biology</i> , 2017, 1492, 127-146.	0.9	13
50	Human intelectin-1 (ITLN1) genetic variation and intestinal expression. <i>Scientific Reports</i> , 2021, 11, 12889.	3.3	13
51	Immunocytochemical detection of ERG expression in exfoliated urinary cells identifies with high specificity patients with prostate cancer. <i>BJU International</i> , 2016, 117, 686-696.	2.5	12
52	A Comparison of Assays for Accurate Copy Number Measurement of the Low-Affinity Fc Gamma Receptor Genes FCGR3A and FCGR3B. <i>PLoS ONE</i> , 2015, 10, e0116791.	2.5	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. <i>PLoS ONE</i> , 2014, 9, e84192.	2.5	11
54	Expansion of a 12-kb VNTR containing the REXO1L1 gene cluster underlies the microscopically visible euchromatic variant of 8q21.2. <i>European Journal of Human Genetics</i> , 2014, 22, 458-463.	2.8	10

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55	Copy number variation of scavenger-receptor cysteine-rich domains within DMBT1 and Crohn's disease. <i>European Journal of Human Genetics</i> , 2016, 24, 1294-1300.	2.8	10
56	Human CCL3L1 copy number variation, gene expression, and the role of the CCL3L1-CCR5 axis in lung function. <i>Wellcome Open Research</i> , 2018, 3, 13.	1.8	10
57	Human intelectin 2 (ITLN2) is selectively expressed by secretory Paneth cells. <i>FASEB Journal</i> , 2022, 36, e22200.	0.5	10
58	Detection of Copy Number Changes in DNA from Formalin Fixed Paraffin Embedded Tissues Using Parologue Ratio Tests. <i>Analytical Chemistry</i> , 2011, 83, 3484-3492.	6.5	9
59	Cohort Profile: Extended Cohort for E-health, Environment and DNA (EXCEED). <i>International Journal of Epidemiology</i> , 2019, 48, 678-679j.	1.9	9
60	Automated design of parologue ratio test assays for the accurate and rapid typing of copy number variation. <i>Bioinformatics</i> , 2013, 29, 1997-2003.	4.1	8
61	Population genetics of immune-related multilocus copy number variation in Native Americans. <i>Journal of the Royal Society Interface</i> , 2017, 14, 20170057.	3.4	8
62	Structural variation of the malaria-associated human glycoporphin A-B-E region. <i>BMC Genomics</i> , 2020, 21, 446.	2.8	7
63	No evidence for DNA copy number change associated with the DUP25 cytogenetic phenotype. <i>European Journal of Human Genetics</i> , 2003, 11, 911-912.	2.8	6
64	Extensive variation in the intelectin gene family in laboratory and wild mouse strains. <i>Scientific Reports</i> , 2021, 11, 15548.	3.3	6
65	Exome-wide analysis of copy number variation shows association of the human leukocyte antigen region with asthma in UK Biobank. <i>BMC Medical Genomics</i> , 2022, 15, .	1.5	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. <i>Clinical and Translational Medicine</i> , 2021, 11, e477.	4.0	5
68	Recurrent mutation at the classical haptoglobin structural polymorphism. <i>Nature Genetics</i> , 2016, 48, 347-348.	21.4	4
69	No Evidence for Association of Defensin Genomic Copy Number with HIV Susceptibility, HIV Load during Clinical Latency, or Progression to AIDS. <i>Annals of Human Genetics</i> , 2017, 81, 27-34.	0.8	4
70	Balancing selection at the human salivary agglutinin gene (DMBT1) driven by host-microbe interactions. <i>IScience</i> , 2022, 25, 104189.	4.1	4
71	Determining multiallelic complex copy number and sequence variation from high coverage exome sequencing data. <i>BMC Genomics</i> , 2015, 16, 891.	2.8	3
72	Analysis of copy number variation at DMBT1 and age-related macular degeneration. <i>BMC Medical Genetics</i> , 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. <i>Haematologica</i> , 2019, 104, e527-e529.	3.5	2
74	Î²-Defensin Genomic Copy Number Does Not Influence the Age of Onset in Huntington's Disease. <i>Journal of Huntington's Disease</i> , 2013, 2, 107-124.	1.9	1
75	Copy number variation of the REXO1L1 gene cluster; euchromatic deletion variant or susceptibility factor?. <i>European Journal of Human Genetics</i> , 2017, 25, 8-9.	2.8	1
76	Genotyping complex structural variation at the malaria-associated human glycoporphin locus using a PCR-based strategy. <i>Annals of Human Genetics</i> , 2021, 85, 7-17.	0.8	1
77	Human CCL3L1 copy number variation, gene expression, and the role of the CCL3L1-CCR5 axis in lung function. <i>Wellcome Open Research</i> , 0, 3, 13.	1.8	1