John A L Armour

List of Publications by Year in descending order

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100 papers	7,214 citations	71102 41 h-index	56724 83 g-index
101 all docs	101 docs citations	101 times ranked	6789 citing authors

#	Article	IF	Citations
1	Psoriasis is associated with increased Î²-defensin genomic copy number. Nature Genetics, 2008, 40, 23-25.	21.4	587
2	Complex gene conversion events in germline mutation at human minisatellites. Nature Genetics, 1994, 6, 136-145.	21.4	524
3	Deletion of the late cornified envelope LCE3B and LCE3C genes as a susceptibility factor for psoriasis. Nature Genetics, 2009, 41, 211-215.	21.4	482
4	Isolation of human simple repeat loci by hybridization selection. Human Molecular Genetics, 1994, 3, 599-605.	2.9	438
5	Uniparental paternal disomy in Angelman's syndrome. Lancet, The, 1991, 337, 694-697.	13.7	330
6	Extensive Normal Copy Number Variation of a Î²-Defensin Antimicrobial-Gene Cluster. American Journal of Human Genetics, 2003, 73, 591-600.	6.2	315
7	The Frequency of Uniparental Disomy in Prader-Willi Syndrome. New England Journal of Medicine, 1992, 326, 1599-1607.	27.0	257
8	Measurement of locus copy number by hybridisation with amplifiable probes. Nucleic Acids Research, 2000, 28, 605-609.	14.5	178
9	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
10	Minisatellite diversity supports a recent African origin for modern humans. Nature Genetics, 1996, 13, 154-160.	21.4	173
11	Genomic copy number variation, human health, and disease. Lancet, The, 2009, 374, 340-350.	13.7	172
12	Systematic cloning of human minisatellites from ordered array charomid libraries. Genomics, 1990, 8, 501-512.	2.9	153
13	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
14	Pendred syndrome (goitre and sensorineural hearing loss) maps to chromosome 7 in the region containing the nonsyndromic deafness gene DFNB4. Nature Genetics, 1996, 12, 421-423.	21.4	146
15	Evolutionary Conservation of a Coding Function for D4Z4, the Tandem DNA Repeat Mutated in Facioscapulohumeral Muscular Dystrophy. American Journal of Human Genetics, 2007, 81, 264-279.	6.2	142
16	Î±-Cardiac myosin heavy chain (MYH6) mutations affecting myofibril formation are associated with congenital heart defects. Human Molecular Genetics, 2010, 19, 4007-4016.	2.9	131
17	Multilocus genetic models of handedness closely resemble singleâ€locus models in explaining family data and are compatible with genomeâ€wide association studies. Annals of the New York Academy of Sciences, 2013, 1288, 48-58.	3.8	129
18	Accurate, high-throughput typing of copy number variation using paralogue ratios from dispersed repeats. Nucleic Acids Research, 2007, 35, e19-e19.	14.5	128

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19	Sequences flanking the repeat arrays of human minlsatellites: association with tandem and dispersed repeat elements. Nucleic Acids Research, 1989, 17, 4925-4936.	14.5	117
20	Analysis of somatic mutations at human minisatellite loci in tumors and cell lines. Genomics, 1989, 4, 328-334.	2.9	113
21	The detection of large deletions or duplications in genomic DNA. Human Mutation, 2002, 20, 325-337.	2.5	110
22	Obesity, starch digestion and amylase: association between copy number variants at human salivary (AMY1) and pancreatic (AMY2) amylase genes. Human Molecular Genetics, 2015, 24, 3472-3480.	2.9	105
23	Genome-wide association study of handedness excludes simple genetic models. Heredity, 2014, 112, 221-225.	2.6	101
24	Allelic diversity at minisatellite MS205 (D16S309): evidence for polarized variability. Human Molecular Genetics, 1993, 2, 1137-1145.	2.9	98
25	Meta-Analysis Confirms the LCE3C_LCE3B Deletion as a Risk Factor for Psoriasis in Several Ethnic Groups and Finds Interaction with HLA-Cw6. Journal of Investigative Dermatology, 2011, 131, 1105-1109.	0.7	89
26	Measurement methods and accuracy in copy number variation: failure to replicate associations of beta-defensin copy number with Crohn's disease. Human Molecular Genetics, 2010, 19, 4930-4938.	2.9	81
27	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
28	Mutation rate heterogeneity and the generation of allele diversity at the human minisatellite MS205 (D16S309). Human Molecular Genetics, 1996, 5, 1823-1833.	2.9	77
29	Experimental aspects of copy number variant assays at CCL3L1. Nature Medicine, 2009, 15, 1115-1117.	30.7	69
30	Screening for subtelomeric chromosome abnormalities in children with idiopathic mental retardation using multiprobe telomeric FISH and the new MAPH telomeric assay. European Journal of Human Genetics, 2001, 9, 527-532.	2.8	67
31	Selective sweep on human amylase genes postdates the split with Neanderthals. Scientific Reports, 2016, 6, 37198.	3.3	67
32	Biology and applications of human minisatellite loci. Current Opinion in Genetics and Development, 1992, 2, 850-856.	3.3	64
33	Mutation processes at human minisatellites. Electrophoresis, 1995, 16, 1577-1585.	2.4	62
34	Replication of LCE3Câ€“LCE3B CNV as a Risk Factor for Psoriasis and Analysis of Interaction with Other Genetic Risk Factors. Journal of Investigative Dermatology, 2010, 130, 979-984.	0.7	61
35	The tetranucleotide repeat polymorphism D21S1245 demonstrates hypermutability in germline and somatic cells. Human Molecular Genetics, 1995, 4, 1193-1199.	2.9	59
36	Hypervariable minisatellite DNA sequences in the Indian peafowl Pavo cristatus. Genomics, 1991, 9, 587-597.	2.9	58

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37	Directional and balancing selection in human beta-defensins. BMC Evolutionary Biology, 2008, 8, 113.	3.2	58
38	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
39	Association of Î²-Defensin Copy Number and Psoriasis in Three Cohorts of European Origin. Journal of Investigative Dermatology, 2012, 132, 2407-2413.	0.7	50
40	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
41	Heritability of Attractiveness to Mosquitoes. PLoS ONE, 2015, 10, e0122716.	2.5	46
42	Multiplex Paralogue Ratio Tests for accurate measurement of multiallelic CNVs. Genomics, 2009, 93, 98-103.	2.9	43
43	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
44	Accuracy and differential bias in copy number measurement of CCL3L1 in association studies with three auto-immune disorders. BMC Genomics, 2011, 12, 418.	2.8	35
45	Low Î±-defensin gene copy number increases the risk for IgA nephropathy and renal dysfunction. Science Translational Medicine, 2016, 8, 345ra88.	12.4	35
46	Copy number variation of human AMY1 is a minor contributor to variation in salivary amylase expression and activity. Human Genomics, 2017, 11, 2.	2.9	35
47	Tandemly repeated DNA: Why should anyone care?. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2006, 598, 6-14.	1.0	32
48	Allelic imbalance on chromosome I in human breast cancer. I. Minisatellite and rflp analysis. Genes Chromosomes and Cancer, 1995, 12, 16-23.	2.8	31
49	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
50	Recurrent Rearrangements of Human Amylase Genes Create Multiple Independent CNV Series. Human Mutation, 2017, 38, 532-539.	2.5	29
51	Direct analysis by small-pool PCR of MS205 minisatellite mutation rates in sperm after mutagenic therapies. Mutation Research - Genetic Toxicology and Environmental Mutagenesis, 1999, 445, 73-80.	1.7	28
52	Constitutional trisomy 8 and BehÃ§et syndrome. American Journal of Medical Genetics, Part A, 2009, 149A, 982-986.	1.2	26
53	Deletion of Late Cornified Envelope 3B and 3C Genes Is Not Associated with Atopic Dermatitis. Journal of Investigative Dermatology, 2010, 130, 2057-2061.	0.7	25
54	CEPH Consortium Map of Chromosome 9. Genomics, 1994, 19, 203-214.	2.9	24

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55	Human minisatellite alleles detectable only after PCR amplification. Genomics, 1992, 12, 116-124.	2.9	23
56	DNA copy number analysis by MAPH: molecular diagnostic applications. Expert Review of Molecular Diagnostics, 2002, 2, 370-378.	3.1	23
57	High-resolution analysis of 16q22.1 in breast carcinoma using DNA amplifiable probes (multiplex) Tj ETQq1 1 0.784314 rgBT /Overloc Cancer, 2005, 114, 720-729.	5.1	22
58	Accurate measurement of gene copy number for human alpha-defensin DEFA1A3. BMC Genomics, 2013, 14, 719.	2.8	22
59	Loss of heterozygosity on the X chromosome in human breast cancer. Genes Chromosomes and Cancer, 1995, 13, 229-238.	2.8	21
60	PLP1 and GPM6B intragenic copy number analysis by MAPH in 262 patients with hypomyelinating leukodystrophies: identification of one partial triplication and two partial deletions of PLP1. Neurogenetics, 2006, 7, 31-37.	1.4	21
61	MS205 Minisatellite Diversity in Basques: Evidence for a Pre-Neolithic Component. Genome Research, 1998, 8, 1289-1298.	5.5	21
62	A hypervariable locus D16S309 located at the distal end of 16p. Nucleic Acids Research, 1992, 20, 1164-1164.	14.5	20
63	CCL3L1 copy number and susceptibility to malaria. Infection, Genetics and Evolution, 2012, 12, 1147-1154.	2.3	20
64	CCL3L1 copy number, CCR5genotype and susceptibility to tuberculosis. BMC Medical Genetics, 2014, 15, 5.	2.1	19
65	Distribution of tandem repeat polymorphism within minisatellite MS621 (D5S110). Annals of Human Genetics, 1996, 60, 11-20.	0.8	18
66	Golli-MBP Copy Number Analysis by FISH, QMPSF and MAPH in 195 Patients with Hypomyelinating Leukodystrophies. Annals of Human Genetics, 2006, 70, 66-77.	0.8	16
67	Integrated analysis of sequence evolution and population history using hypervariable compound haplotypes. Human Molecular Genetics, 2000, 9, 2675-2681.	2.9	14
68	Evolution and population genetics of the H-ras minisatellite and cancer predisposition. Human Molecular Genetics, 2003, 12, 891-900.	2.9	14
69	Thyroid peroxidase: evidence for disease gene exclusion in Pendred's syndrome. Clinical Endocrinology, 1996, 44, 441-446.	2.4	13
70	Functional effects of CCL3L1 copy number. Genes and Immunity, 2012, 13, 374-379.	4.1	13
71	No Evidence for Association of BMI with Salivary Amylase Gene Copy Number in the UK 1958 Birth Cohort. Obesity, 2019, 27, 1533-1538.	3.0	13
72	Cis-regulation of inter-allelic exchanges in mutation at human minisatellite MS205 in yeast. Gene, 1999, 232, 143-153.	2.2	12

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73	Microarray MAPH: accurate array-based detection of relative copy number in genomic DNA. BMC Genomics, 2006, 7, 163.	2.8	11
74	Isolation of human minisatellite loci detected by synthetic tandem repeat probes: direct comparison with cloned DNA fingerprinting probes. Human Molecular Genetics, 1992, 1, 319-323.	2.9	10
75	Abnormal Segregation of Alleles in CEPH Pedigree DNAs Arising from Allele Loss in Lymphoblastoid DNA. Genomics, 1993, 15, 119-122.	2.9	10
76	Screening for exonic copy number mutations at MSH2 and MLH1 by MAPH. Familial Cancer, 2005, 4, 145-149.	1.9	10
77	Association analysis of the CCL3L1 copy number locus by paralogue ratio test in Norwegian rheumatoid arthritis patients and healthy controls. Genes and Immunity, 2012, 13, 579-582.	4.1	10
78	Recent advances in minisatellite biology. FEBS Letters, 1992, 307, 113-115.	2.8	9
79	Evaluation of tetranucleotide repeat locus D7S809 (wg1g9) in the Japanese population. Forensic Science International, 1996, 81, 133-140.	2.2	9
80	Skin microbiome alters attractiveness to Anopheles mosquitoes. BMC Microbiology, 2022, 22, 98.	3.3	9
81	Sequence analysis of alleles at a microsatellite locus D14S299 (wg1c5) and population genetic comparisons. International Journal of Legal Medicine, 1999, 113, 15-18.	2.2	8
82	STS for minisatellite MS607 (D22S163). Nucleic Acids Research, 1991, 19, 3158-3158.	14.5	6
83	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
84	Compound haplotypes at Xp11.23 and human population growth in Eurasia. Annals of Human Genetics, 2004, 68, 428-437.	0.8	6
85	Inferring mechanisms of copy number change from haplotype structures at the human DEFA1A3 locus. BMC Genomics, 2014, 15, 614.	2.8	6
86	Evaluation of two new STR loci 9q2h2 and wg3f12 in a Japanese population. Legal Medicine, 1999, 1, 25-28.	1.3	4
87	Tetrameric short tandem repeat (STR) system D15S233 (wg1d1): sequencing and frequency data in the japanese and Chinese populations. Legal Medicine, 1999, 1, 119-126.	1.3	4
88	Copy number variation and antigenic repertoire. Nature Genetics, 2009, 41, 1263-1264.	21.4	4
89	Determination of haplotypes at structurally complex regions using emulsion haplotype fusion PCR. BMC Genomics, 2012, 13, 693.	2.8	4
90	Quadruplex MAPH: improvement of throughput in high-resolution copy number screening. BMC Genomics, 2009, 10, 453.	2.8	3

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91	Sharp focus on the variable genome. Nature, 2009, 461, 735-736.	27.8	3
92	Gene Dosage Analysis by Multiplex Amplifiable Probe Hybridization. , 2004, 92, 125-140.		3
93	Screening for common copy-number variants in cancer genes. Cancer Genetics and Cytogenetics, 2010, 203, 316-323.	1.0	2
94	Evolution of haplotypes at CCL3L1/CCL4L1. Genome Biology, 2010, 11, .	8.8	1
95	Analysis of Multiallelic CNVs by Emulsion Haplotype Fusion PCR. Methods in Molecular Biology, 2017, 1492, 155-165.	0.9	1
96	A New Triplex STR System Without Irregular Alleles by Silver Staining and Its Potential Application to Forensic Analysis. Journal of Forensic Sciences, 2001, 46, 448-452.	1.6	1
97	33.6 (D1S111) and pMLAJ1 (D1S61) identify the same VNTR on chromosome 1. Nucleic Acids Research, 1991, 19, 4801-4801.	14.5	0
98	STS for minisatellite 33.1 (D9S49): direct typing by PCR. Nucleic Acids Research, 1991, 19, 4788-4788.	14.5	0
99	Human Genetics: Measuring the Raw Material of Evolution. Current Biology, 2009, 19, R736-R738.	3.9	0
100	Recombination and Human Diversity. Stadler Genetics Symposia Series, 2000, , 81-89.	0.0	0