Andrew J Sharp

List of Publications by Year
in descending order

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86	9,413	71102	58581
papers	citations	41 h-index	82 g-index

97	97	97	13140
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Fine-scale structural variation of the human genome. Nature Genetics, 2005, 37, 727-732.	21.4	897
2	Segmental Duplications and Copy-Number Variation in the Human Genome. American Journal of Human Genetics, 2005, 77, 78-88.	6.2	872
3	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. New England Journal of Medicine, 2008, 359, 1685-1699.	27.0	663
4	Discovery of previously unidentified genomic disorders from the duplication architecture of the human genome. Nature Genetics, 2006, 38, 1038-1042.	21.4	557
5	15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. Nature Genetics, 2009, 41, 160-162.	21.4	511
6	A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. Nature Genetics, 2008, 40, 322-328.	21.4	509
7	Age- and tissue-specific variation of X chromosome inactivation ratios in normal women. Human Genetics, 2000, 107, 343-349.	3.8	309
8	Abundant contribution of short tandem repeats to gene expression variation in humans. Nature Genetics, 2016, 48, 22-29.	21.4	291
9	Linkage Disequilibrium and Heritability of Copy-Number Polymorphisms within Duplicated Regions of the Human Genome. American Journal of Human Genetics, 2006, 79, 275-290.	6.2	283
10	Structural Variation of the Human Genome. Annual Review of Genomics and Human Genetics, 2006, 7, 407-442.	6.2	255
11	DNA methylation profiles of human active and inactive X chromosomes. Genome Research, 2011, 21, 1592-1600.	5.5	244
12	Epigenome-wide differences in pathology-free regions of multiple sclerosisâ€“affected brains. Nature Neuroscience, 2014, 17, 121-130.	14.8	239
13	Recurrent Reciprocal Genomic Rearrangements of 17q12 Are Associated with Renal Disease, Diabetes, and Epilepsy. American Journal of Human Genetics, 2007, 81, 1057-1069.	6.2	222
14	Identification of the translocation breakpoints in the Ts65Dn and Ts1Cje mouse lines: relevance for modeling down syndrome. Mammalian Genome, 2011, 22, 674-684.	2.2	186
15	Characterization of a recurrent 15q24 microdeletion syndrome. Human Molecular Genetics, 2007, 16, 567-572.	2.9	173
16	Genome-wide DNA methylation profiling in the superior temporal gyrus reveals epigenetic signatures associated with Alzheimerâ€™s disease. Genome Medicine, 2016, 8, 5.	8.2	163
17	Xp deletions associated with autism in three females. Human Genetics, 1999, 104, 43-48.	3.8	150
18	The Genetics of Microdeletion and Microduplication Syndromes: An Update. Annual Review of Genomics and Human Genetics, 2014, 15, 215-244.	6.2	145

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19	Genome-Wide DNA Methylation Profiling Reveals Epigenetic Changes in the Rat Nucleus Accumbens Associated With Cross-Generational Effects of Adolescent THC Exposure. Neuropsychopharmacology, 2015, 40, 2993-3005.	5.4	143
20	DNA Methylation Signatures of Early Childhood Malnutrition Associated With Impairments in Attention and Cognition. Biological Psychiatry, 2016, 80, 765-774.	1.3	124
21	Polymorphic tandem repeats within gene promoters act as modifiers of gene expression and DNA methylation in humans. Nucleic Acids Research, 2016, 44, 3750-3762.	14.5	120
22	Dynamics of DNA Methylation in Recent Human and Great Ape Evolution. PLoS Genetics, 2013, 9, e1003763.	3.5	118
23	Molecular and cytogenetic analysis of the spreading of X inactivation in X;autosome translocations. Human Molecular Genetics, 2002, 11, 3145-3156.	2.9	90
24	DNA Methylation: Insights into Human Evolution. PLoS Genetics, 2015, 11, e1005661.	3.5	90
25	Effect of Copy Number Variants on Outcomes for Infants With Single Ventricle Heart Defects. Circulation: Cardiovascular Genetics, 2013, 6, 444-451.	5.1	89
26	Identification of rare de novo epigenetic variations in congenital disorders. Nature Communications, 2018, 9, 2064.	12.8	82
27	Emerging themes and new challenges in defining the role of structural variation in human disease. Human Mutation, 2009, 30, 135-144.	2.5	79
28	Placental expression profile of imprinted genes impacts birth weight. Epigenetics, 2015, 10, 842-849.	2.7	79
29	A meta-analysis of epigenome-wide association studies in Alzheimerâ€™s disease highlights novel differentially methylated loci across cortex. Nature Communications, 2021, 12, 3517.	12.8	72
30	Tandem repeat variation in human and great ape populations and its impact on gene expression divergence. Genome Research, 2015, 25, 1591-1599.	5.5	69
31	Foxa2 identifies a cardiac progenitor population with ventricular differentiation potential. Nature Communications, 2017, 8, 14428.	12.8	68
32	The interplay between DNA methylation and sequence divergence in recent human evolution. Nucleic Acids Research, 2015, 43, 8204-8214.	14.5	67
33	DNA Methylation Profiling of Uniparental Disomy Subjects Provides a Map of Parental Epigenetic Bias in the Human Genome. American Journal of Human Genetics, 2016, 99, 555-566.	6.2	66
34	The complex SNP and CNV genetic architecture of the increased risk of congenital heart defects in Down syndrome. Genome Research, 2013, 23, 1410-1421.	5.5	65
35	A survey of inter-individual variation in DNA methylation identifies environmentally responsive co-regulated networks of epigenetic variation in the human genome. PLoS Genetics, 2018, 14, e1007707.	3.5	65
36	A study of females with deletions of the short arm of the X chromosome. Human Genetics, 1998, 102, 507-516.	3.8	64

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37	Parent of origin effects. Clinical Genetics, 2012, 81, 201-209.	2.0	59
38	Expression of imprinted genes in placenta is associated with infant neurobehavioral development. Epigenetics, 2015, 10, 834-841.	2.7	59
39	A Novel Framework for Characterizing Genomic Haplotype Diversity in the Human Immunoglobulin Heavy Chain Locus. Frontiers in Immunology, 2020, 11, 2136.	4.8	54
40	RNA analysis reveals splicing mutations and loss of expression defects inMLH1 andBRCA1. Human Mutation, 2004, 24, 272-272.	2.5	52
41	Loss of RNA expression and allele-specific expression associated with congenital heart disease. Nature Communications, 2016, 7, 12824.	12.8	51
42	Absence of correlation between late-replication and spreading of X inactivation in an X;autosome translocation. Human Genetics, 2001, 109, 295-302.	3.8	49
43	Digital Genotyping of Macrosatellites and Multicopy Genes Reveals Novel Biological Functions Associated with Copy Number Variation of Large Tandem Repeats. PLoS Genetics, 2014, 10, e1004418.	3.5	49
44	Back to the past in schizophrenia genomics. Nature Neuroscience, 2016, 19, 1-2.	14.8	49
45	Dual transcriptomic and epigenomic study of reaction severity in peanut-allergic children. Journal of Allergy and Clinical Immunology, 2020, 145, 1219-1230.	2.9	44
46	DNA methylation profiling in X;autosome translocations supports a role for L1 repeats in the spread of X chromosome inactivation. Human Molecular Genetics, 2014, 23, 1224-1236.	2.9	43
47	Methylation profiling in individuals with uniparental disomy identifies novel differentially methylated regions on chromosome 15. Genome Research, 2010, 20, 1271-1278.	5.5	42
48	A Survey of Rare Epigenetic Variation in 23,116 Human Genomes Identifies Disease-Relevant Epivariations and CGG Expansions. American Journal of Human Genetics, 2020, 107, 654-669.	6.2	40
49	Opposite Phenotypes of Muscle Strength and Locomotor Function in Mouse Models of Partial Trisomy and Monosomy 21 for the Proximal Hspa13-App Region. PLoS Genetics, 2015, 11, e1005062.	3.5	39
50	A recurrent 14q32.2 microdeletion mediated by expanded TGG repeats. Human Molecular Genetics, 2010, 19, 1967-1973.	2.9	36
51	The telomeric part of the human chromosome 21 from Cstb to Prmt2 is not necessary for the locomotor and short-term memory deficits observed in the Tc1 mouse model of Down syndrome. Behavioural Brain Research, 2011, 217, 271-281.	2.2	34
52	Tandem repeat sequence variation as causative Cis-eQTLs for protein-coding gene expression variation: The case of CSTB. Human Mutation, 2012, 33, 1302-1309.	2.5	34
53	Episignatures Stratifying Helsmoortel-Van Der Aa Syndrome Show Modest Correlation with Phenotype. American Journal of Human Genetics, 2020, 107, 555-563.	6.2	32
54	Pervasive cis effects of variation in copy number of large tandem repeats on local DNA methylation and gene expression. American Journal of Human Genetics, 2021, 108, 809-824.	6.2	30

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55	High-throughput genotyping of intermediate-size structural variation. Human Molecular Genetics, 2006, 15, 1159-1167.	2.9	28
56	Rapid Multiplexed Genotyping of Simple Tandem Repeats using Capture and High-Throughput Sequencing. Human Mutation, 2013, 34, 1304-1311.	2.5	28
57	Rare genetic variation at transcription factor binding sites modulates local DNA methylation profiles. PLoS Genetics, 2020, 16, e1009189.	3.5	27
58	Optimal design of oligonucleotide microarrays for measurement of DNA copy-number. Human Molecular Genetics, 2007, 16, 2770-2779.	2.9	25
59	The App-Runx1 Region Is Critical for Birth Defects and Electrocardiographic Dysfunctions Observed in a Down Syndrome Mouse Model. PLoS Genetics, 2012, 8, e1002724.	3.5	25
60	RNA-Seq in 296 phased trios provides a high-resolution map of genomic imprinting. BMC Biology, 2019, 17, 50.	3.8	23
61	MsPAC: a tool for haplotype-phased structural variant detection. Bioinformatics, 2020, 36, 922-924.	4.1	23
62	Fumarates target the metabolic-epigenetic interplay of brain-homing T cells in multiple sclerosis. Brain, 2019, 142, 647-661.	7.6	22
63	A 3-way hybrid approach to generate a new high-quality chimpanzee reference genome (Pan_tro_3.0). GigaScience, 2017, 6, 1-6.	6.4	17
64	Clinical Characterization of Copy Number Variants Associated With Neurodevelopmental Disorders in a Large-scale Multiancestry Biobank. JAMA Psychiatry, 2022, 79, 250.	11.0	16
65	Evaluation of PRDM9 variation as a risk factor for recurrent genomic disorders and chromosomal non-disjunction. Human Genetics, 2012, 131, 1519-1524.	3.8	15
66	Screening for rare epigenetic variations in autism and schizophrenia. Human Mutation, 2019, 40, 952-961.	2.5	14
67	Gene Dosage Analysis in Silver-Russell Syndrome: Use of Quantitative Competitive PCR and Dual-Color FISH to Estimate the Frequency of Duplications in 7p11.2â€“p13. Genetic Testing and Molecular Biomarkers, 2001, 5, 261-266.	1.7	12
68	Genome-wide analysis of parent-of-origin effects in non-syndromic orofacial clefts. European Journal of Human Genetics, 2014, 22, 822-830.	2.8	12
69	A phenome-wide association study identifies effects of copy-number variation of VNTRs and multicopy genes on multiple human traits. American Journal of Human Genetics, 2022, 109, 1065-1076.	6.2	12
70	Detection of Genomic Variation by Selection of a 9 Mb DNA Region and High Throughput Sequencing. PLoS ONE, 2009, 4, e6659.	2.5	11
71	Detection of Parent-of-Origin Specific Expression Quantitative Trait Loci by Cis-Association Analysis of Gene Expression in Trios. PLoS ONE, 2012, 7, e41695.	2.5	11
72	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

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73	Potockiâ€“Lupski syndrome mimicking a connective tissue disorder. Clinical Dysmorphology, 2008, 17, 211-213.	0.3	9
74	Genome-wide linkage and copy number variation analysis reveals 710â€…kb duplication on chromosome 1p31.3 responsible for autosomal dominant omphalocele. Journal of Medical Genetics, 2012, 49, 270-276.	3.2	9
75	Evidence from skewed X inactivation for trisomy mosaicism in Silver-Russell syndrome. European Journal of Human Genetics, 2001, 9, 887-891.	2.8	8
76	Limitations of lymphoblastoid cell lines for establishing genetic reference datasets in the immunoglobulin loci. PLoS ONE, 2021, 16, e0261374.	2.5	4
77	Comment on â€œGenomic Hypomethylation in the Human Germline Associates with Selective Structural Mutability in the Human Genomeâ€. PLoS Genetics, 2013, 9, e1003332.	3.5	3
78	Determining multiallelic complex copy number and sequence variation from high coverage exome sequencing data. BMC Genomics, 2015, 16, 891.	2.8	3
79	Robust identification of deletions in exome and genome sequence data based on clustering of Mendelian errors. Human Mutation, 2018, 39, 870-881.	2.5	3
80	Elucidation of de novo small insertion/deletion biology with parentâ€ofâ€origin phasing. Human Mutation, 2020, 41, 800-806.	2.5	3
81	Genome-wide DNA methylation analysis in patients with familial ATR-X mental retardation syndrome. , 0, , 434-446.		1
82	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
83	The Effects of Childhood Malnutrition on DNA Methylation in Adulthood. FASEB Journal, 2015, 29, 749.2.	0.5	1
84	Angels in Marble: Working Class Conservatives in Urban England, by Robert Mc Kenzie and Allan Silver, Heinemann, London, 1969, 285 pp., $6.20.. Political Science, 1969, 21, 54-57.	0.6	0
85	Revealing the hidden structure of our genome. Nature Methods, 2006, 3, 427-428.	19.0	0
86	Whole Genome Methylation Profiling by Immunoprecipitation of Methylated DNA. Methods in Molecular Biology, 2012, 925, 69-78.	0.9	0