

# Andrew J Sharp

## List of Publications by Year in descending order

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86  
papers

9,413  
citations

70961

41  
h-index

58464

82  
g-index

97  
all docs

97  
docs citations

97  
times ranked

13140  
citing authors

#	ARTICLE	IF	CITATIONS
1	Fine-scale structural variation of the human genome. <i>Nature Genetics</i> , 2005, 37, 727-732.	9.4	897
2	Segmental Duplications and Copy-Number Variation in the Human Genome. <i>American Journal of Human Genetics</i> , 2005, 77, 78-88.	2.6	872
3	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. <i>New England Journal of Medicine</i> , 2008, 359, 1685-1699.	13.9	663
4	Discovery of previously unidentified genomic disorders from the duplication architecture of the human genome. <i>Nature Genetics</i> , 2006, 38, 1038-1042.	9.4	557
5	15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. <i>Nature Genetics</i> , 2009, 41, 160-162.	9.4	511
6	A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. <i>Nature Genetics</i> , 2008, 40, 322-328.	9.4	509
7	Age- and tissue-specific variation of X chromosome inactivation ratios in normal women. <i>Human Genetics</i> , 2000, 107, 343-349.	1.8	309
8	Abundant contribution of short tandem repeats to gene expression variation in humans. <i>Nature Genetics</i> , 2016, 48, 22-29.	9.4	291
9	Linkage Disequilibrium and Heritability of Copy-Number Polymorphisms within Duplicated Regions of the Human Genome. <i>American Journal of Human Genetics</i> , 2006, 79, 275-290.	2.6	283
10	Structural Variation of the Human Genome. <i>Annual Review of Genomics and Human Genetics</i> , 2006, 7, 407-442.	2.5	255
11	DNA methylation profiles of human active and inactive X chromosomes. <i>Genome Research</i> , 2011, 21, 1592-1600.	2.4	244
12	Epigenome-wide differences in pathology-free regions of multiple sclerosis-affected brains. <i>Nature Neuroscience</i> , 2014, 17, 121-130.	7.1	239
13	Recurrent Reciprocal Genomic Rearrangements of 17q12 Are Associated with Renal Disease, Diabetes, and Epilepsy. <i>American Journal of Human Genetics</i> , 2007, 81, 1057-1069.	2.6	222
14	Identification of the translocation breakpoints in the Ts65Dn and Ts1Cje mouse lines: relevance for modeling down syndrome. <i>Mammalian Genome</i> , 2011, 22, 674-684.	1.0	186
15	Characterization of a recurrent 15q24 microdeletion syndrome. <i>Human Molecular Genetics</i> , 2007, 16, 567-572.	1.4	173
16	Genome-wide DNA methylation profiling in the superior temporal gyrus reveals epigenetic signatures associated with Alzheimer's disease. <i>Genome Medicine</i> , 2016, 8, 5.	3.6	163
17	Xp deletions associated with autism in three females. <i>Human Genetics</i> , 1999, 104, 43-48.	1.8	150
18	The Genetics of Microdeletion and Microduplication Syndromes: An Update. <i>Annual Review of Genomics and Human Genetics</i> , 2014, 15, 215-244.	2.5	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. <i>Neuropsychopharmacology</i> , 2015, 40, 2993-3005.	2.8	143
20	DNA Methylation Signatures of Early Childhood Malnutrition Associated With Impairments in Attention and Cognition. <i>Biological Psychiatry</i> , 2016, 80, 765-774.	0.7	124
21	Polymorphic tandem repeats within gene promoters act as modifiers of gene expression and DNA methylation in humans. <i>Nucleic Acids Research</i> , 2016, 44, 3750-3762.	6.5	120
22	Dynamics of DNA Methylation in Recent Human and Great Ape Evolution. <i>PLoS Genetics</i> , 2013, 9, e1003763.	1.5	118
23	Molecular and cytogenetic analysis of the spreading of X inactivation in X;autosome translocations. <i>Human Molecular Genetics</i> , 2002, 11, 3145-3156.	1.4	90
24	DNA Methylation: Insights into Human Evolution. <i>PLoS Genetics</i> , 2015, 11, e1005661.	1.5	90
25	Effect of Copy Number Variants on Outcomes for Infants With Single Ventricle Heart Defects. <i>Circulation: Cardiovascular Genetics</i> , 2013, 6, 444-451.	5.1	89
26	Identification of rare de novo epigenetic variations in congenital disorders. <i>Nature Communications</i> , 2018, 9, 2064.	5.8	82
27	Emerging themes and new challenges in defining the role of structural variation in human disease. <i>Human Mutation</i> , 2009, 30, 135-144.	1.1	79
28	Placental expression profile of imprinted genes impacts birth weight. <i>Epigenetics</i> , 2015, 10, 842-849.	1.3	79
29	A meta-analysis of epigenome-wide association studies in Alzheimer's disease highlights novel differentially methylated loci across cortex. <i>Nature Communications</i> , 2021, 12, 3517.	5.8	72
30	Tandem repeat variation in human and great ape populations and its impact on gene expression divergence. <i>Genome Research</i> , 2015, 25, 1591-1599.	2.4	69
31	Foxa2 identifies a cardiac progenitor population with ventricular differentiation potential. <i>Nature Communications</i> , 2017, 8, 14428.	5.8	68
32	The interplay between DNA methylation and sequence divergence in recent human evolution. <i>Nucleic Acids Research</i> , 2015, 43, 8204-8214.	6.5	67
33	DNA Methylation Profiling of Uniparental Disomy Subjects Provides a Map of Parental Epigenetic Bias in the Human Genome. <i>American Journal of Human Genetics</i> , 2016, 99, 555-566.	2.6	66
34	The complex SNP and CNV genetic architecture of the increased risk of congenital heart defects in Down syndrome. <i>Genome Research</i> , 2013, 23, 1410-1421.	2.4	65
35	A survey of inter-individual variation in DNA methylation identifies environmentally responsive co-regulated networks of epigenetic variation in the human genome. <i>PLoS Genetics</i> , 2018, 14, e1007707.	1.5	65
36	A study of females with deletions of the short arm of the X chromosome. <i>Human Genetics</i> , 1998, 102, 507-516.	1.8	64

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37	Parent of origin effects. <i>Clinical Genetics</i> , 2012, 81, 201-209.	1.0	59
38	Expression of imprinted genes in placenta is associated with infant neurobehavioral development. <i>Epigenetics</i> , 2015, 10, 834-841.	1.3	59
39	A Novel Framework for Characterizing Genomic Haplotype Diversity in the Human Immunoglobulin Heavy Chain Locus. <i>Frontiers in Immunology</i> , 2020, 11, 2136.	2.2	54
40	RNA analysis reveals splicing mutations and loss of expression defects in MLH1 and BRCA1. <i>Human Mutation</i> , 2004, 24, 272-272.	1.1	52
41	Loss of RNA expression and allele-specific expression associated with congenital heart disease. <i>Nature Communications</i> , 2016, 7, 12824.	5.8	51
42	Absence of correlation between late-replication and spreading of X inactivation in an X;autosome translocation. <i>Human Genetics</i> , 2001, 109, 295-302.	1.8	49
43	Digital Genotyping of Macrosatellites and Multicopy Genes Reveals Novel Biological Functions Associated with Copy Number Variation of Large Tandem Repeats. <i>PLoS Genetics</i> , 2014, 10, e1004418.	1.5	49
44	Back to the past in schizophrenia genomics. <i>Nature Neuroscience</i> , 2016, 19, 1-2.	7.1	49
45	Dual transcriptomic and epigenomic study of reaction severity in peanut-allergic children. <i>Journal of Allergy and Clinical Immunology</i> , 2020, 145, 1219-1230.	1.5	44
46	DNA methylation profiling in X;autosome translocations supports a role for L1 repeats in the spread of X chromosome inactivation. <i>Human Molecular Genetics</i> , 2014, 23, 1224-1236.	1.4	43
47	Methylation profiling in individuals with uniparental disomy identifies novel differentially methylated regions on chromosome 15. <i>Genome Research</i> , 2010, 20, 1271-1278.	2.4	42
48	A Survey of Rare Epigenetic Variation in 23,116 Human Genomes Identifies Disease-Relevant Epivariations and CGG Expansions. <i>American Journal of Human Genetics</i> , 2020, 107, 654-669.	2.6	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. <i>PLoS Genetics</i> , 2015, 11, e1005062.	1.5	39
50	A recurrent 14q32.2 microdeletion mediated by expanded TCG repeats. <i>Human Molecular Genetics</i> , 2010, 19, 1967-1973.	1.4	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. <i>Behavioural Brain Research</i> , 2011, 217, 271-281.	1.2	34
52	Tandem repeat sequence variation as causative Cis-eQTLs for protein-coding gene expression variation: The case of CSTB. <i>Human Mutation</i> , 2012, 33, 1302-1309.	1.1	34
53	Episignatures Stratifying Helsmoortel-Van Der Aa Syndrome Show Modest Correlation with Phenotype. <i>American Journal of Human Genetics</i> , 2020, 107, 555-563.	2.6	32
54	Pervasive cis effects of variation in copy number of large tandem repeats on local DNA methylation and gene expression. <i>American Journal of Human Genetics</i> , 2021, 108, 809-824.	2.6	30

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55	High-throughput genotyping of intermediate-size structural variation. <i>Human Molecular Genetics</i> , 2006, 15, 1159-1167.	1.4	28
56	Rapid Multiplexed Genotyping of Simple Tandem Repeats using Capture and High-Throughput Sequencing. <i>Human Mutation</i> , 2013, 34, 1304-1311.	1.1	28
57	Rare genetic variation at transcription factor binding sites modulates local DNA methylation profiles. <i>PLoS Genetics</i> , 2020, 16, e1009189.	1.5	27
58	Optimal design of oligonucleotide microarrays for measurement of DNA copy-number. <i>Human Molecular Genetics</i> , 2007, 16, 2770-2779.	1.4	25
59	The App-Runx1 Region Is Critical for Birth Defects and Electrocardiographic Dysfunctions Observed in a Down Syndrome Mouse Model. <i>PLoS Genetics</i> , 2012, 8, e1002724.	1.5	25
60	RNA-Seq in 296 phased trios provides a high-resolution map of genomic imprinting. <i>BMC Biology</i> , 2019, 17, 50.	1.7	23
61	MsPAC: a tool for haplotype-phased structural variant detection. <i>Bioinformatics</i> , 2020, 36, 922-924.	1.8	23
62	Fumarates target the metabolic-epigenetic interplay of brain-homing T cells in multiple sclerosis. <i>Brain</i> , 2019, 142, 647-661.	3.7	22
63	A 3-way hybrid approach to generate a new high-quality chimpanzee reference genome (Pan_tro_3.0). <i>GigaScience</i> , 2017, 6, 1-6.	3.3	17
64	Clinical Characterization of Copy Number Variants Associated With Neurodevelopmental Disorders in a Large-scale Multiancestry Biobank. <i>JAMA Psychiatry</i> , 2022, 79, 250.	6.0	16
65	Evaluation of PRDM9 variation as a risk factor for recurrent genomic disorders and chromosomal non-disjunction. <i>Human Genetics</i> , 2012, 131, 1519-1524.	1.8	15
66	Screening for rare epigenetic variations in autism and schizophrenia. <i>Human Mutation</i> , 2019, 40, 952-961.	1.1	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. <i>Genetic Testing and Molecular Biomarkers</i> , 2001, 5, 261-266.	1.7	12
68	Genome-wide analysis of parent-of-origin effects in non-syndromic orofacial clefts. <i>European Journal of Human Genetics</i> , 2014, 22, 822-830.	1.4	12
69	A phenome-wide association study identifies effects of copy-number variation of VNTRs and multicopy genes on multiple human traits. <i>American Journal of Human Genetics</i> , 2022, 109, 1065-1076.	2.6	12
70	Detection of Genomic Variation by Selection of a 9 Mb DNA Region and High Throughput Sequencing. <i>PLoS ONE</i> , 2009, 4, e6659.	1.1	11
71	Detection of Parent-of-Origin Specific Expression Quantitative Trait Loci by Cis-Association Analysis of Gene Expression in Trios. <i>PLoS ONE</i> , 2012, 7, e41695.	1.1	11
72	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.	1.4	10

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73	Potockiâ€“Lupski syndrome mimicking a connective tissue disorder. <i>Clinical Dysmorphology</i> , 2008, 17, 211-213.	0.1	9
74	Genome-wide linkage and copy number variation analysis reveals 710â€“kb duplication on chromosome 1p31.3 responsible for autosomal dominant omphalocele. <i>Journal of Medical Genetics</i> , 2012, 49, 270-276.	1.5	9
75	Evidence from skewed X inactivation for trisomy mosaicism in Silver-Russell syndrome. <i>European Journal of Human Genetics</i> , 2001, 9, 887-891.	1.4	8
76	Limitations of lymphoblastoid cell lines for establishing genetic reference datasets in the immunoglobulin loci. <i>PLoS ONE</i> , 2021, 16, e0261374.	1.1	4
77	Comment on â€œGenomic Hypomethylation in the Human Germline Associates with Selective Structural Mutability in the Human Genomeâ€: <i>PLoS Genetics</i> , 2013, 9, e1003332.	1.5	3
78	Determining multiallelic complex copy number and sequence variation from high coverage exome sequencing data. <i>BMC Genomics</i> , 2015, 16, 891.	1.2	3
79	Robust identification of deletions in exome and genome sequence data based on clustering of Mendelian errors. <i>Human Mutation</i> , 2018, 39, 870-881.	1.1	3
80	Elucidation of de novo small insertion/deletion biology with parentâ€“origin phasing. <i>Human Mutation</i> , 2020, 41, 800-806.	1.1	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?. <i>European Journal of Human Genetics</i> , 2017, 25, 8-9.	1.4	1
83	The Effects of Childhood Malnutrition on DNA Methylation in Adulthood. <i>FASEB Journal</i> , 2015, 29, 749.2.	0.2	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.. <i>Political Science</i> , 1969, 21, 54-57.	0.3	0
85	Revealing the hidden structure of our genome. <i>Nature Methods</i> , 2006, 3, 427-428.	9.0	0
86	Whole Genome Methylation Profiling by Immunoprecipitation of Methylated DNA. <i>Methods in Molecular Biology</i> , 2012, 925, 69-78.	0.4	0