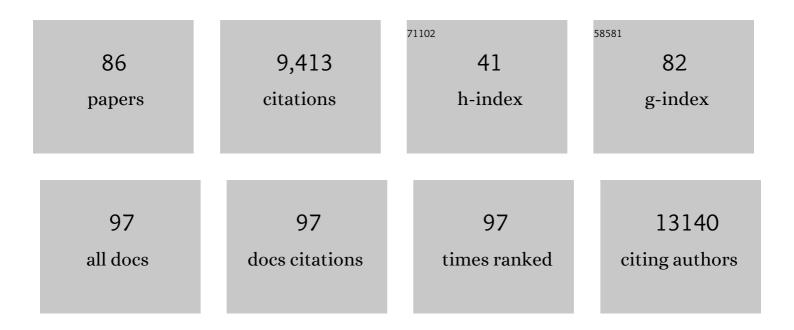
Andrew J Sharp

List of Publications by Year in descending order

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| # | Article | IF | CITATIONS |
|----|---|------|-----------|
| 1 | Clinical Characterization of Copy Number Variants Associated With Neurodevelopmental Disorders in a Large-scale Multiancestry Biobank. JAMA Psychiatry, 2022, 79, 250. | 11.0 | 16 |
| 2 | 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 |
| 3 | 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 |
| 4 | A meta-analysis of epigenome-wide association studies in Alzheimer's disease highlights novel differentially methylated loci across cortex. Nature Communications, 2021, 12, 3517. | 12.8 | 72 |
| 5 | Limitations of lymphoblastoid cell lines for establishing genetic reference datasets in the immunoglobulin loci. PLoS ONE, 2021, 16, e0261374. | 2.5 | 4 |
| 6 | MsPAC: a tool for haplotype-phased structural variant detection. Bioinformatics, 2020, 36, 922-924. | 4.1 | 23 |
| 7 | Elucidation of de novo small insertion/deletion biology with parentâ€ofâ€origin phasing. Human Mutation, 2020, 41, 800-806. | 2.5 | 3 |
| 8 | 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 |
| 9 | 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 |
| 10 | A Novel Framework for Characterizing Genomic Haplotype Diversity in the Human Immunoglobulin Heavy Chain Locus. Frontiers in Immunology, 2020, 11, 2136. | 4.8 | 54 |
| 11 | Episignatures Stratifying Helsmoortel-Van Der Aa Syndrome Show Modest Correlation with Phenotype. American Journal of Human Genetics, 2020, 107, 555-563. | 6.2 | 32 |
| 12 | Rare genetic variation at transcription factor binding sites modulates local DNA methylation profiles. PLoS Genetics, 2020, 16, e1009189. | 3.5 | 27 |
| 13 | RNA-Seq in 296 phased trios provides a high-resolution map of genomic imprinting. BMC Biology, 2019, 17, 50. | 3.8 | 23 |
| 14 | Fumarates target the metabolic-epigenetic interplay of brain-homing T cells in multiple sclerosis. Brain, 2019, 142, 647-661. | 7.6 | 22 |
| 15 | Screening for rare epigenetic variations in autism and schizophrenia. Human Mutation, 2019, 40, 952-961. | 2.5 | 14 |
| 16 | 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 |
| 17 | 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 |
| 18 | Identification of rare de novo epigenetic variations in congenital disorders. Nature Communications, 2018, 9, 2064 | 12.8 | 82 |

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|----|--|------|-----------|
| 19 | Foxa2 identifies a cardiac progenitor population with ventricular differentiation potential. Nature Communications, 2017, 8, 14428. | 12.8 | 68 |
| 20 | 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 |
| 21 | 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 |
| 22 | 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 |
| 23 | 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 |
| 24 | Loss of RNA expression and allele-specific expression associated with congenital heart disease. Nature Communications, 2016, 7, 12824. | 12.8 | 51 |
| 25 | Genome-wide DNA methylation profiling in the superior temporal gyrus reveals epigenetic signatures associated with Alzheimer's disease. Genome Medicine, 2016, 8, 5. | 8.2 | 163 |
| 26 | Abundant contribution of short tandem repeats to gene expression variation in humans. Nature Genetics, 2016, 48, 22-29. | 21.4 | 291 |
| 27 | DNA Methylation Signatures of Early Childhood Malnutrition Associated With Impairments in Attention and Cognition. Biological Psychiatry, 2016, 80, 765-774. | 1.3 | 124 |
| 28 | Back to the past in schizophrenia genomics. Nature Neuroscience, 2016, 19, 1-2. | 14.8 | 49 |
| 29 | Determining multiallelic complex copy number and sequence variation from high coverage exome sequencing data. BMC Genomics, 2015, 16, 891. | 2.8 | 3 |
| 30 | DNA Methylation: Insights into Human Evolution. PLoS Genetics, 2015, 11, e1005661. | 3.5 | 90 |
| 31 | 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 |
| 32 | Placental expression profile of imprinted genes impacts birth weight. Epigenetics, 2015, 10, 842-849. | 2.7 | 79 |
| 33 | Expression of imprinted genes in placenta is associated with infant neurobehavioral development. Epigenetics, 2015, 10, 834-841. | 2.7 | 59 |
| 34 | 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 |
| 35 | The interplay between DNA methylation and sequence divergence in recent human evolution. Nucleic Acids Research, 2015, 43, 8204-8214. | 14.5 | 67 |
| 36 | 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 |

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|----|--|------|-----------|
| 37 | The Effects of Childhood Malnutrition on DNA Methylation in Adulthood. FASEB Journal, 2015, 29, 749.2. | 0.5 | 1 |
| 38 | 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 |
| 39 | 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 |
| 40 | 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 |
| 41 | Epigenome-wide differences in pathology-free regions of multiple sclerosis–affected brains. Nature Neuroscience, 2014, 17, 121-130. | 14.8 | 239 |
| 42 | 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 |
| 43 | The Genetics of Microdeletion and Microduplication Syndromes: An Update. Annual Review of Genomics and Human Genetics, 2014, 15, 215-244. | 6.2 | 145 |
| 44 | Rapid Multiplexed Genotyping of Simple Tandem Repeats using Capture and High-Throughput Sequencing. Human Mutation, 2013, 34, 1304-1311. | 2.5 | 28 |
| 45 | Comment on "Genomic Hypomethylation in the Human Germline Associates with Selective Structural Mutability in the Human Genome― PLoS Genetics, 2013, 9, e1003332. | 3.5 | 3 |
| 46 | Dynamics of DNA Methylation in Recent Human and Great Ape Evolution. PLoS Genetics, 2013, 9, e1003763. | 3.5 | 118 |
| 47 | Effect of Copy Number Variants on Outcomes for Infants With Single Ventricle Heart Defects. Circulation: Cardiovascular Genetics, 2013, 6, 444-451. | 5.1 | 89 |
| 48 | 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 |
| 49 | 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 |
| 50 | 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 |
| 51 | 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 |
| 52 | Whole Genome Methylation Profiling by Immunoprecipitation of Methylated DNA. Methods in Molecular Biology, 2012, 925, 69-78. | 0.9 | 0 |
| 53 | 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 |
| 54 | 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 |

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|----|--|------|-----------|
| 55 | Parent of origin effects. Clinical Genetics, 2012, 81, 201-209. | 2.0 | 59 |
| 56 | 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 |
| 57 | ldentification 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 |
| 58 | DNA methylation profiles of human active and inactive X chromosomes. Genome Research, 2011, 21, 1592-1600. | 5.5 | 244 |
| 59 | Methylation profiling in individuals with uniparental disomy identifies novel differentially methylated regions on chromosome 15. Genome Research, 2010, 20, 1271-1278. | 5.5 | 42 |
| 60 | A recurrent 14q32.2 microdeletion mediated by expanded TGG repeats. Human Molecular Genetics, 2010, 19, 1967-1973. | 2.9 | 36 |
| 61 | Detection of Genomic Variation by Selection of a 9 Mb DNA Region and High Throughput Sequencing. PLoS ONE, 2009, 4, e6659. | 2.5 | 11 |
| 62 | Emerging themes and new challenges in defining the role of structural variation in human disease. Human Mutation, 2009, 30, 135-144. | 2.5 | 79 |
| 63 | 15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. Nature Genetics, 2009, 41, 160-162. | 21.4 | 511 |
| 64 | A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. Nature Genetics, 2008, 40, 322-328. | 21.4 | 509 |
| 65 | Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. New England Journal of Medicine, 2008, 359, 1685-1699. | 27.0 | 663 |
| 66 | Potocki–Lupski syndrome mimicking a connective tissue disorder. Clinical Dysmorphology, 2008, 17, 211-213. | 0.3 | 9 |
| 67 | Characterization of a recurrent 15q24 microdeletion syndrome. Human Molecular Genetics, 2007, 16, 567-572. | 2.9 | 173 |
| 68 | Optimal design of oligonucleotide microarrays for measurement of DNA copy-number. Human Molecular Genetics, 2007, 16, 2770-2779. | 2.9 | 25 |
| 69 | 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 |
| 70 | 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 |
| 71 | Structural Variation of the Human Genome. Annual Review of Genomics and Human Genetics, 2006, 7, 407-442. | 6.2 | 255 |
| 72 | Discovery of previously unidentified genomic disorders from the duplication architecture of the human genome. Nature Genetics, 2006, 38, 1038-1042. | 21.4 | 557 |

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|----|---|------|-----------|
| 73 | Revealing the hidden structure of our genome. Nature Methods, 2006, 3, 427-428. | 19.0 | Ο |
| 74 | High-throughput genotyping of intermediate-size structural variation. Human Molecular Genetics, 2006, 15, 1159-1167. | 2.9 | 28 |
| 75 | Fine-scale structural variation of the human genome. Nature Genetics, 2005, 37, 727-732. | 21.4 | 897 |
| 76 | Segmental Duplications and Copy-Number Variation in the Human Genome. American Journal of Human Genetics, 2005, 77, 78-88. | 6.2 | 872 |
| 77 | RNA analysis reveals splicing mutations and loss of expression defects inMLH1 andBRCA1. Human Mutation, 2004, 24, 272-272. | 2.5 | 52 |
| 78 | Molecular and cytogenetic analysis of the spreading of X inactivation in X;autosome translocations. Human Molecular Genetics, 2002, 11, 3145-3156. | 2.9 | 90 |
| 79 | 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 |
| 80 | Evidence from skewed X inactivation for trisomy mosaicism in Silver-Russell syndrome. European Journal of Human Genetics, 2001, 9, 887-891. | 2.8 | 8 |
| 81 | Gene Dosage Analysis in Silver-Russell Syndrome: Use of Quantitative Competitive PCR and Dual-Color FISH to Estimate the Frequency of Duplications in 7p11.2–p13. Genetic Testing and Molecular Biomarkers, 2001, 5, 261-266. | 1.7 | 12 |
| 82 | Age- and tissue-specific variation of X chromosome inactivation ratios in normal women. Human Genetics, 2000, 107, 343-349. | 3.8 | 309 |
| 83 | Xp deletions associated with autism in three females. Human Genetics, 1999, 104, 43-48. | 3.8 | 150 |
| 84 | A study of females with deletions of the short arm of the X chromosome. Human Genetics, 1998, 102, 507-516. | 3.8 | 64 |
| 85 | 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 | Ο |
| 86 | Genome-wide DNA methylation analysis in patients with familial ATR-X mental retardation syndrome. , 0, , 434-446. | | 1 |