Michael A Eberle

List of Publications by Year
in descending order

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Version: 2024-02-01

48 papers	35,337 citations	101496 36 h-index	197736 49 g-index
65 all docs	65 docs citations	65 times ranked	56571 citing authors

#	Article	IF	Citations
1	Whole genome sequencing for the diagnosis of neurological repeat expansion disorders in the UK: a retrospective diagnostic accuracy and prospective clinical validation study. Lancet Neurology, The, 2022, 21, 234-245.	4.9	74
2	The Effect of <scp>SMN</scp> Gene Dosage on <scp>ALS</scp> Risk and Disease Severity. Annals of Neurology, 2021, 89, 686-697.	2.8	10
3	Cyrius: accurate CYP2D6 genotyping using whole-genome sequencing data. Pharmacogenomics Journal, 2021, 21, 251-261.	0.9	50
4	Intronic Haplotypes in the <scp>GBA</scp> Gene Do Not Predict Age at Diagnosis of Parkinson's Disease. Movement Disorders, 2021, 36, 1456-1460.	2.2	5
5	Genome-wide sequencing as a first-tier screening test for short tandem repeat expansions. Genome Medicine, 2021, 13, 126.	3.6	27
6	Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. Nature Genetics, 2021, 53, 1636-1648.	9.4	223
7	Repeat expansions confer WRN dependence in microsatellite-unstable cancers. Nature, 2020, 586, 292-298.	13.7	95
8	Large scale in silico characterization of repeat expansion variation in human genomes. Scientific Data, 2020, 7, 294.	2.4	12
9	Genome-wide detection of tandem DNA repeats that are expanded in autism. Nature, 2020, 586, 80-86.	13.7	155
10	Spinal muscular atrophy diagnosis and carrier screening from genome sequencing data. Genetics in Medicine, 2020, 22, 945-953.	1.1	78
11	ExpansionHunter Denovo: a computational method for locating known and novel repeat expansions in short-read sequencing data. Genome Biology, 2020, 21, 102.	3.8	114
12	Bioinformatics-Based Identification of Expanded Repeats: A Non-reference Intronic Pentamer Expansion in RFC1 Causes CANVAS. American Journal of Human Genetics, 2019, 105, 151-165.	2.6	170
13	ExpansionHunter: a sequence-graph-based tool to analyze variation in short tandem repeat regions. Bioinformatics, 2019, 35, 4754-4756.	1.8	183
14	Length of Uninterrupted CAG, Independent of Polyglutamine Size, Results in Increased Somatic Instability, Hastening Onset of Huntington Disease. American Journal of Human Genetics, 2019, 104, 1116-1126.	2.6	130
15	Best practices for benchmarking germline small-variant calls in human genomes. Nature Biotechnology, 2019, 37, 555-560.	9.4	273
16	Glutaminase Deficiency Caused by Short Tandem Repeat Expansion in GLS. New England Journal of Medicine, 2019, 380, 1433-1441.	13.9	71
17	Paragraph: a graph-based structural variant genotyper for short-read sequence data. Genome Biology, 2019, 20, 291.	3.8	104
18	Copy-number variants in clinical genome sequencing: deployment and interpretation for rare and undiagnosed disease. Genetics in Medicine, 2019, 21, 1121-1130.	1.1	83

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19	A reference data set of 5.4 million phased human variants validated by genetic inheritance from sequencing a three-generation 17-member pedigree. Genome Research, 2017, 27, 157-164.	2.4	338
20	Detection of long repeat expansions from PCR-free whole-genome sequence data. Genome Research, 2017, 27, 1895-1903.	2.4	277
21	A global reference for human genetic variation. Nature, 2015, 526, 68-74.	13.7	13,998
22	Whole-genome haplotyping by dilution, amplification, and sequencing. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 5552-5557.	3.3	67
23	Improved imputation of common and uncommon SNPs with a new reference set. Nature Genetics, 2012, 44, 6-7.	9.4	45
24	An integrated map of genetic variation from 1,092 human genomes. Nature, 2012, 491, 56-65.	13.7	7,199
25	A map of human genome variation from population-scale sequencing. Nature, 2010, 467, 1061-1073.	13.7	7,209
26	Power to Detect Risk Alleles Using Genome-Wide Tag SNP Panels. PLoS Genetics, 2007, 3, e170.	1.5	89
27	Sequence diversity, natural selection and linkage disequilibrium in the human T cell receptor alpha/delta locus. Human Genetics, 2006, 119, 255-266.	1.8	19
28	Allele Frequency Matching Between SNPs Reveals an Excess of Linkage Disequilibrium in Genic Regions of the Human Genome. PLoS Genetics, 2006, 2, e142.	1.5	72
29	TRPV6 exhibits unusual patterns of polymorphism and divergence in worldwide populations. Human Molecular Genetics, 2006, 15, 2106-2113.	1.4	58
30	COMMENT ON 'DISCREPANCIES IN dbSNP CONFIRMATIONS RATES AND ALLELE FREQUENCY DISTRIBUTIONS FROM VARYING GENOTYPING ERROR RATES AND PATTERNS'. Bioinformatics, 2005, 21, 141-143.	1.8	6
31	Genomic regions exhibiting positive selection identified from dense genotype data. Genome Research, 2005, 15, 1553-1565.	2.4	235
32	Population History and Natural Selection Shape Patterns of Genetic Variation in 132 Genes. PLoS Biology, 2004, 2, e286.	2.6	455
33	Mapping complex disease loci in whole-genome association studies. Nature, 2004, 429, 446-452.	13.7	580
34	Extensive and breed-specific linkage disequilibrium in Canis familiaris. Genome Research, 2004, 14, 2388-2396.	2.4	273
35	Selecting a Maximally Informative Set of Single-Nucleotide Polymorphisms for Association Analyses Using Linkage Disequilibrium. American Journal of Human Genetics, 2004, 74, 106-120.	2.6	1,469
36	Haplotype Diversity across 100 Candidate Genes for Inflammation, Lipid Metabolism, and Blood Pressure Regulation in Two Populations. American Journal of Human Genetics, 2004, 74, 610-622.	2.6	163

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37	Additional SNPs and linkage-disequilibrium analyses are necessary for whole-genome association studies in humans. Nature Genetics, 2003, 33, 518-521.	9.4	299
38	A numerical study of the interaction between the mantle wedge, subducting slab, and overriding plate. Physics of the Earth and Planetary Interiors, 2002, 134, 191-202.	0.7	79
39	A New Susceptibility Locus for Autosomal Dominant Pancreatic Cancer Maps to Chromosome 4q32-34. American Journal of Human Genetics, 2002, 70, 1044-1048.	2.6	123
40	Sequence variation in the human T-cell receptor loci. Immunological Reviews, 2002, 190, 26-39.	2.8	26
41	Sequence Variation and Linkage Disequilibrium in the Human T-Cell Receptor Î² (TCRB) Locus. American Journal of Human Genetics, 2001, 69, 381-395.	2.6	54
42	Lower-Than-Expected Linkage Disequilibrium between Tightly Linked Markers in Humans Suggests a Role for Gene Conversion. American Journal of Human Genetics, 2001, 69, 582-589.	2.6	95
43	An analysis of strategies for discovery of single-nucleotide polymorphisms. Genetic Epidemiology, 2000, 19, S29-S35.	0.6	43
44	Sampling SNPs. Nature Genetics, 2000, 26, 13-14.	9.4	56
45	Evidence from the asymmetry of fast-spreading ridges that the axial topographic high is due to extensional stresses. Nature, 1998, 394, 360-363.	13.7	18
46	Constraints on a buoyant model for the formation of the axial topographic high on the East Pacific Rise. Journal of Geophysical Research, 1998, 103, 12291-12307.	3.3	15
47	An alternative, dynamic model of the axial topographic high at fast spreading ridges. Journal of Geophysical Research, 1998, 103, 12309-12320.	3.3	30
48	Regional viscosity variations, small-scale convection and slope of the depth-age1/2curve. Geophysical Research Letters, 1995, 22, 473-476.	1.5	15