David J Cutler

List of Publications by Year
in descending order

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98	17,050	87888 38	42399 92
papers	citations	h-index	g-index

111	111	111	27506
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A second generation human haplotype map of over 3.1 million SNPs. Nature, 2007, 449, 851-861.	27.8	4,137
2	Synaptic, transcriptional and chromatin genes disrupted in autism. Nature, 2014, 515, 209-215.	27.8	2,254
3	Genome-wide detection and characterization of positive selection in human populations. Nature, 2007, 449, 913-918.	27.8	1,788
4	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. Cell, 2020, 180, 568-584.e23.	28.9	1,422
5	A Note on Exact Tests of Hardy-Weinberg Equilibrium. American Journal of Human Genetics, 2005, 76, 887-893.	6.2	1,232
6	Genomic alterations in cultured human embryonic stem cells. Nature Genetics, 2005, 37, 1099-1103.	21.4	592
7	A Common Genetic Variant in the Neurexin Superfamily Member CNTNAP2 Increases Familial Risk of Autism. American Journal of Human Genetics, 2008, 82, 160-164.	6.2	566
8	A common sex-dependent mutation in a RET enhancer underlies Hirschsprung disease risk. Nature, 2005, 434, 857-863.	27.8	438
9	Microarray-based genomic selection for high-throughput resequencing. Nature Methods, 2007, 4, 907-909.	19.0	374
10	Refining the role of de novo protein-truncating variants in neurodevelopmental disorders by using population reference samples. Nature Genetics, 2017, 49, 504-510.	21.4	298
11	High-Throughput Variation Detection and Genotyping Using Microarrays. Genome Research, 2001, 11, 1913-1925.	5.5	258
12	Microdeletions of 3q29 Confer High Risk for Schizophrenia. American Journal of Human Genetics, 2010, 87, 229-236.	6.2	215
13	Autosomal Recessive Causes Likely in Early-Onset Alzheimer Disease. Archives of Neurology, 2012, 69, 59.	4.5	193
14	Haplotype Inference in Random Population Samples. American Journal of Human Genetics, 2002, 71, 1129-1137.	6.2	176
15	Undetected Genotyping Errors Cause Apparent Overtransmission of Common Alleles in the Transmission/Disequilibrium Test. American Journal of Human Genetics, 2003, 72, 598-610.	6.2	157
16	Exhaustive allelic transmission disequilibrium tests as a new approach to genome-wide association studies. Nature Genetics, 2004, 36, 1181-1188.	21.4	154
17	Genome-Wide Association Study Identifies African-Specific Susceptibility Loci in African Americans With Inflammatory Bowel Disease. Gastroenterology, 2017, 152, 206-217.e2.	1.3	120
18	Recessive gene disruptions in autism spectrum disorder. Nature Genetics, 2019, 51, 1092-1098.	21.4	109

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19	Identification of novel FMR1 variants by massively parallel sequencing in developmentally delayed males. American Journal of Medical Genetics, Part A, 2010, 152A, 2512-2520.	1.2	108
20	Tracking the Evolution of the SARS Coronavirus Using High-Throughput, High-Density Resequencing Arrays. Genome Research, 2004, 14, 398-405.	5.5	104
21	Association of Early-Onset Alzheimer Disease With Elevated Low-Density Lipoprotein Cholesterol Levels and Rare Genetic Coding Variants of APOB. JAMA Neurology, 2019, 76, 809.	9.0	94
22	Blood-Derived DNA Methylation Signatures of Crohn's Disease and Severity of Intestinal Inflammation. Gastroenterology, 2019, 156, 2254-2265.e3.	1.3	91
23	Estimating Divergence Times in the Presence of an Overdispersed Molecular Clock. Molecular Biology and Evolution, 2000, 17, 1647-1660.	8.9	88
24	Genetic contributors to risk of schizophrenia in the presence of a 22q11.2 deletion. Molecular Psychiatry, 2021, 26, 4496-4510.	7.9	87
25	To Pool, or Not to Pool?. Genetics, 2010, 186, 41-43.	2.9	86
26	PATTERNS OFGENETICVARIATION INMENDELIAN ANDCOMPLEXTRAITS. Annual Review of Genomics and Human Genetics, 2000, 1, 387-407.	6.2	78
27	A Statistical Approach for Testing Cross-Phenotype Effects of Rare Variants. American Journal of Human Genetics, 2016, 98, 525-540.	6.2	75
28	Reciprocal Duplication of the Williams-Beuren Syndrome Deletion on Chromosome 7q11.23 Is Associated with Schizophrenia. Biological Psychiatry, 2014, 75, 371-377.	1.3	66
29	An Oligonucleotide Microarray for High-Throughput Sequencing of the Mitochondrial Genome. Journal of Molecular Diagnostics, 2006, 8, 476-482.	2.8	65
30	Characterization of Genetic Loci That Affect Susceptibility to Inflammatory Bowel Diseases in African Americans. Gastroenterology, 2015, 149, 1575-1586.	1.3	65
31	Microarray-based resequencing of multiple Bacillus anthracis isolates. Genome Biology, 2004, 6, R10.	9.6	64
32	Understanding the Overdispersed Molecular Clock. Genetics, 2000, 154, 1403-1417.	2.9	64
33	Clinical and Genomic Correlates of Neutrophil Reactive Oxygen Species Production in Pediatric Patients With Crohnâ€™s Disease. Gastroenterology, 2018, 154, 2097-2110.	1.3	63
34	Identification of Polycystic Kidney Disease 1 Like 1 Gene Variants in Children With Biliary Atresia Splenic Malformation Syndrome. Hepatology, 2019, 70, 899-910.	7.3	58
35	Population Bottlenecks as a Potential Major Shaping Force of Human Genome Architecture. PLoS Genetics, 2007, 3, e119.	3.5	55
36	Discrepancies in dbSNP confirmation rates and allele frequency distributions from varying genotyping error rates and patterns. Bioinformatics, 2004, 20, 1022-1032.	4.1	52

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37	Genetic control of the human brain proteome. American Journal of Human Genetics, 2021, 108, 400-410.	6.2	52
38	Integrating Next-Generation Genomic Sequencing and Mass Spectrometry To Estimate Allele-Specific Protein Abundance in Human Brain. Journal of Proteome Research, 2017, 16, 3336-3347.	3.7	48
39	Genome-wide Enrichment of De Novo Coding Mutations in Orofacial Cleft Trios. American Journal of Human Genetics, 2020, 107, 124-136.	6.2	48
40	Exome Sequencing Identifies a Novel FOXP3 Mutation in a 2â€Generation Family With Inflammatory Bowel Disease. Journal of Pediatric Gastroenterology and Nutrition, 2014, 58, 561-568.	1.8	47
41	Haplotype and Missing Data Inference in Nuclear Families. Genome Research, 2004, 14, 1624-1632.	5.5	42
42	SeqAnt: A web service to rapidly identify and annotate DNA sequence variations. BMC Bioinformatics, 2010, 11, 471.	2.6	38
43	Dissecting Allele Architecture of Early Onset IBD Using High-Density Genotyping. PLoS ONE, 2015, 10, e0128074.	2.5	35
44	Excess variants in AFF2 detected by massively parallel sequencing of males with autism spectrum disorder. Human Molecular Genetics, 2012, 21, 4356-4364.	2.9	34
45	Population Demographic History Can Cause the Appearance of Recombination Hotspots. American Journal of Human Genetics, 2012, 90, 774-783.	6.2	33
46	PEMapper and PECaller provide a simplified approach to whole-genome sequencing. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E1923-E1932.	7.1	31
47	Validation and extension of an empirical Bayes method for SNP calling on Affymetrix microarrays. Genome Biology, 2008, 9, R63.	9.6	30
48	Prevalence of intellectual disability among eight-year-old children from selected communities in the United States, 2014. Disability and Health Journal, 2021, 14, 101023.	2.8	30
49	Bystro: rapid online variant annotation and natural-language filtering at whole-genome scale. Genome Biology, 2018, 19, 14.	8.8	29
50	Genome-Wide Association Study of Down Syndrome-Associated Atrioventricular Septal Defects. G3: Genes, Genomes, Genetics, 2015, 5, 1961-1971.	1.8	28
51	Array-Based FMR1 Sequencing and Deletion Analysis in Patients with a Fragile X Syndromeâ€“Like Phenotype. PLoS ONE, 2010, 5, e9476.	2.5	26
52	Multiplex Chromosomal Exome Sequencing Accelerates Identification of ENU-Induced Mutations in the Mouse. G3: Genes, Genomes, Genetics, 2012, 2, 143-150.	1.8	25
53	Progress and Disparities in Early Identification of Autism Spectrum Disorder: Autism and Developmental Disabilities Monitoring Network, 2002-2016. Journal of the American Academy of Child and Adolescent Psychiatry, 2022, 61, 905-914.	0.5	25
54	On the probability that a novel variant is a disease-causing mutation. Genome Research, 2005, 15, 960-966.	5.5	24

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55	Variation in the Lymphotoxin-Î±/Tumor Necrosis Factor Locus Modifies Risk of Erythema Nodosum in Sarcoidosis. Journal of Investigative Dermatology, 2009, 129, 1921-1926.	0.7	24
56	Contribution of copy-number variation to Down syndromeâ€“associated atrioventricular septal defects. Genetics in Medicine, 2015, 17, 554-560.	2.4	24
57	Rigor of nonâ€dairy galactose restriction in early childhood, measured by retrospective survey, does not associate with severity of five longâ€term outcomes quantified in 231 children and adults with classic galactosemia. Journal of Inherited Metabolic Disease, 2017, 40, 813-821.	3.6	23
58	MPD: multiplex primer design for next-generation targeted sequencing. BMC Bioinformatics, 2017, 18, 14.	2.6	23
59	Enhanced Contribution of HLA in Pediatric Onset Ulcerative Colitis. Inflammatory Bowel Diseases, 2018, 24, 829-838.	1.9	23
60	Targeted sequencing of the human X chromosome exome. Genomics, 2011, 98, 260-265.	2.9	22
61	Identification of rare X-linked neuroligin variants by massively parallel sequencing in males with autism spectrum disorder. Molecular Autism, 2012, 3, 8.	4.9	22
62	Genome-Wide Association Study to Find Modifiers for Tetralogy of Fallot in the 22q11.2 Deletion Syndrome Identifies Variants in the GPR98 Locus on 5q14.3. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	22
63	Genetic variants and pathways implicated in a pediatric inflammatory bowel disease cohort. Genes and Immunity, 2019, 20, 131-142.	4.1	22
64	Whole-genome sequencing of African Americans implicates differential genetic architecture in inflammatory bowel disease. American Journal of Human Genetics, 2021, 108, 431-445.	6.2	21
65	Classic Weinstein: Tetrad Analysis, Genetic Variation and Achiasmate Segregation in Drosophila and Humans. Genetics, 1999, 152, 1615-1629.	2.9	21
66	Common NOD2 risk variants in African Americans with CrohnÊ¼s disease are due exclusively to recent Caucasian admixture. Inflammatory Bowel Diseases, 2012, 18, 2357-2359.	1.9	18
67	Simultaneous Discovery and Testing of Deletions for Disease Association in SNP Genotyping Studies. American Journal of Human Genetics, 2007, 81, 684-699.	6.2	17
68	Population Genetics Identifies Challenges in Analyzing Rare Variants. Genetic Epidemiology, 2015, 39, 145-148.	1.3	16
69	Empirical Evaluation of Oligonucleotide Probe Selection for DNA Microarrays. PLoS ONE, 2010, 5, e9921.	2.5	14
70	Identifying genetic factors that contribute to the increased risk of congenital heart defects in infants with Down syndrome. Scientific Reports, 2020, 10, 18051.	3.3	14
71	Ileal Derived Organoids From Crohnâ€™s Disease Patients Show Unique Transcriptomic and Secretomic Signatures. Cellular and Molecular Gastroenterology and Hepatology, 2021, 12, 1267-1280.	4.5	14
72	Site- and Taxa-Specific Disease-Associated Oral Microbial Structures Distinguish Inflammatory Bowel Diseases. Inflammatory Bowel Diseases, 2021, 27, 1889-1900.	1.9	14

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73	Analysis of Copy Number Variants on Chromosome 21 in Down Syndrome-Associated Congenital Heart Defects. G3: Genes, Genomes, Genetics, 2018, 8, 105-111.	1.8	13
74	Testing Two Evolutionary Theories of Human Aging with DNA Methylation Data. Genetics, 2017, 207, 1547-1560.	2.9	12
75	Eicosatetraynoic Acid and Butyrate Regulate Human Intestinal Organoid Mitochondrial and Extracellular Matrix Pathways Implicated in Crohnâ€™s Disease Strictures. Inflammatory Bowel Diseases, 2022, 28, 988-1003.	1.9	12
76	Investigating the role ofp11 (S100A10) sequence variation in susceptibility to major depression. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2007, 144B, 1079-1082.	1.7	11
77	Not All Autism Genes Are Created Equal: A Response to Myers etÂal.. American Journal of Human Genetics, 2020, 107, 1000-1003.	6.2	11
78	Profiling non-coding RNA levels with clinical classifiers in pediatric Crohnâ€™s disease. BMC Medical Genomics, 2021, 14, 194.	1.5	11
79	The PAX1 locus at 20p11 is a potential genetic modifier for bilateral cleft lip. Human Genetics and Genomics Advances, 2021, 2, 100025.	1.7	9
80	The Index of Dispersion of Molecular Evolution: Slow Fluctuations. Theoretical Population Biology, 2000, 57, 177-186.	1.1	8
81	Genetic and Transcriptomic Variation Linked to Neutrophil Granulocyteâ€“Macrophage Colony-Stimulating Factor Signaling in Pediatric Crohnâ€™s Disease. Inflammatory Bowel Diseases, 2019, 25, 547-560.	1.9	8
82	LDL cholesterol is associated with higher AD neuropathology burden independent of APOE. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, 930-938.	1.9	8
83	A model of binding on DNA microarrays: understanding the combined effect of probe synthesis failure, cross-hybridization, DNA fragmentation and other experimental details of affymetrix arrays. BMC Genomics, 2012, 13, 737.	2.8	7
84	Identification of PSMB5 as a genetic modifier of fragile Xâ€“associated tremor/ataxia syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	7.1	7
85	Microarray oligonucleotide probe designer: a Web service. Open Access Bioinformatics, 2010, 2, 145.	0.9	5
86	Novel Missense CNTNAP2 Variant Identified in Two Consanguineous Pakistani Families With Developmental Delay, Epilepsy, Intellectual Disability, and Aggressive Behavior. Frontiers in Neurology, 0, 13, .	2.4	5
87	Neutrophil GM-CSF signaling in inflammatory bowel disease patients is influenced by non-coding genetic variants. Scientific Reports, 2019, 9, 9168.	3.3	3
88	Response to Graffelman: Tests of Hardy-Weinberg Equilibrium. American Journal of Human Genetics, 2010, 86, 818-819.	6.2	2
89	Sex-specific recombination patterns predict parent of origin for recurrent genomic disorders. BMC Medical Genomics, 2021, 14, 154.	1.5	2
90	Clustered Mutations Have No Effect on the Overdispersed Molecular Clock: A Response to Huai and Woodruff. Genetics, 1998, 149, 463-464.	2.9	2

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91	Methylation quantitative trait loci are largely consistent across disease states in Crohnâ€™s disease. G3: Genes, Genomes, Genetics, 2022, 12, .	1.8	2
92	Reply to PlÃ¼ss et al.: The strength of PEMapper/PECaller lies in unbiased calling using large sample sizes. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E8323-E8323.	7.1	1
93	SeqAnt. , 2017, , .		1
94	Identifying novel causal genes and proteins in Alzheimerâ€™s disease. Alzheimer's and Dementia, 2020, 16, e043523.	0.8	1
95	Integrating human brain proteomes and genomeâ€wide association results implicates new genes in Alzheimerâ€™s disease. Alzheimer's and Dementia, 2020, 16, e043865.	0.8	1
96	Leveraging Family History in Case-Control Analyses of Rare Variation. Genetics, 2020, 214, 295-303.	2.9	0
97	Salmonellosis Outbreak After a Large-Scale Food Event in Virginia, 2017. Public Health Reports, 2020, 135, 668-675.	2.5	0
98	A Comprehensive Search for Recombinogenic Motifs in the Human Genome. PLoS ONE, 2013, 8, e62920.	2.5	0