David J Cutler

List of Publications by Year in descending order

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98 papers 17,050 citations

38 h-index 92 g-index

111 all docs

111 docs citations

times ranked

111

27506 citing authors

#	Article	IF	CITATIONS
1	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
2	Methylation quantitative trait loci are largely consistent across disease states in Crohn's disease. G3: Genes, Genomes, Genetics, 2022, 12, .	1.8	2
3	Eicosatetraynoic Acid and Butyrate Regulate Human Intestinal Organoid Mitochondrial and Extracellular Matrix Pathways Implicated in Crohn's Disease Strictures. Inflammatory Bowel Diseases, 2022, 28, 988-1003.	1.9	12
4	Identification of ⟨i⟩PSMB5⟨ i⟩ as a genetic modifier of fragile X–associated tremor/ataxia syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	7.1	7
5	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
6	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
7	lleal Derived Organoids From Crohn's Disease Patients Show Unique Transcriptomic and Secretomic Signatures. Cellular and Molecular Gastroenterology and Hepatology, 2021, 12, 1267-1280.	4.5	14
8	Genetic control of the human brain proteome. American Journal of Human Genetics, 2021, 108, 400-410.	6.2	52
9	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
10	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
11	Site- and Taxa-Specific Disease-Associated Oral Microbial Structures Distinguish Inflammatory Bowel Diseases. Inflammatory Bowel Diseases, 2021, 27, 1889-1900.	1.9	14
12	Sex-specific recombination patterns predict parent of origin for recurrent genomic disorders. BMC Medical Genomics, 2021, 14, 154.	1.5	2
13	Profiling non-coding RNA levels with clinical classifiers in pediatric Crohn's disease. BMC Medical Genomics, 2021, 14, 194.	1.5	11
14	Genetic contributors to risk of schizophrenia in the presence of a 22q11.2 deletion. Molecular Psychiatry, 2021, 26, 4496-4510.	7.9	87
15	Leveraging Family History in Case-Control Analyses of Rare Variation. Genetics, 2020, 214, 295-303.	2.9	0
16	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
17	Salmonellosis Outbreak After a Large-Scale Food Event in Virginia, 2017. Public Health Reports, 2020, 135, 668-675.	2.5	O
18	Identifying novel causal genes and proteins in Alzheimer's disease. Alzheimer's and Dementia, 2020, 16, e043523.	0.8	1

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19	Integrating human brain proteomes and genomeâ€wide association results implicates new genes in Alzheimer's disease. Alzheimer's and Dementia, 2020, 16, e043865.	0.8	1
20	Not All Autism Genes Are Created Equal: A Response to Myers etÂal American Journal of Human Genetics, 2020, 107, 1000-1003.	6.2	11
21	Genome-wide Enrichment of De Novo Coding Mutations in Orofacial Cleft Trios. American Journal of Human Genetics, 2020, 107, 124-136.	6.2	48
22	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
23	Genetic and Transcriptomic Variation Linked to Neutrophil Granulocyte–Macrophage Colony-Stimulating Factor Signaling in Pediatric Crohn's Disease. Inflammatory Bowel Diseases, 2019, 25, 547-560.	1.9	8
24	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
25	Recessive gene disruptions in autism spectrum disorder. Nature Genetics, 2019, 51, 1092-1098.	21.4	109
26	Neutrophil GM-CSF signaling in inflammatory bowel disease patients is influenced by non-coding genetic variants. Scientific Reports, 2019, 9, 9168.	3. 3	3
27	Association of Early-Onset Alzheimer Disease With Elevated Low-Density Lipoprotein Cholesterol Levels and Rare Genetic Coding Variants of <i>APOB</i> . JAMA Neurology, 2019, 76, 809.	9.0	94
28	Blood-Derived DNA Methylation Signatures of Crohn's Disease and Severity of Intestinal Inflammation. Gastroenterology, 2019, 156, 2254-2265.e3.	1.3	91
29	Genetic variants and pathways implicated in a pediatric inflammatory bowel disease cohort. Genes and Immunity, 2019, 20, 131-142.	4.1	22
30	Clinical and Genomic Correlates of Neutrophil Reactive Oxygen Species Production in Pediatric Patients With Crohn's Disease. Gastroenterology, 2018, 154, 2097-2110.	1.3	63
31	Enhanced Contribution of HLA in Pediatric Onset Ulcerative Colitis. Inflammatory Bowel Diseases, 2018, 24, 829-838.	1.9	23
32	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
33	Bystro: rapid online variant annotation and natural-language filtering at whole-genome scale. Genome Biology, 2018, 19, 14.	8.8	29
34	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
35	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
36	Genome-Wide Association Study to Find Modifiers for Tetralogy of Fallot in the 22q11.2 Deletion Syndrome Identifies Variants in the $\langle i \rangle$ GPR98 $\langle i \rangle$ Locus on 5q14.3. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	22

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37	Reply to $Pl\tilde{A}^{1}/4$ ss 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
38	Testing Two Evolutionary Theories of Human Aging with DNA Methylation Data. Genetics, 2017, 207, 1547-1560.	2.9	12
39	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
40	Rigor of nonâ€dairy galactose restriction in early childhood, measured by retrospective survey, does not associate with severity of five longâ€ŧerm outcomes quantified in 231 children and adults with classic galactosemia. Journal of Inherited Metabolic Disease, 2017, 40, 813-821.	3 . 6	23
41	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
42	SeqAnt., 2017,,.		1
43	MPD: multiplex primer design for next-generation targeted sequencing. BMC Bioinformatics, 2017, 18, 14.	2.6	23
44	A Statistical Approach for Testing Cross-Phenotype Effects of Rare Variants. American Journal of Human Genetics, 2016, 98, 525-540.	6.2	75
45	Genome-Wide Association Study of Down Syndrome-Associated Atrioventricular Septal Defects. G3: Genes, Genomes, Genetics, 2015, 5, 1961-1971.	1.8	28
46	Dissecting Allele Architecture of Early Onset IBD Using High-Density Genotyping. PLoS ONE, 2015, 10, e0128074.	2.5	35
47	Contribution of copy-number variation to Down syndrome–associated atrioventricular septal defects. Genetics in Medicine, 2015, 17, 554-560.	2.4	24
48	Population Genetics Identifies Challenges in Analyzing Rare Variants. Genetic Epidemiology, 2015, 39, 145-148.	1.3	16
49	Characterization of Genetic Loci That Affect Susceptibility to Inflammatory Bowel Diseases in African Americans. Gastroenterology, 2015, 149, 1575-1586.	1.3	65
50	Exome Sequencing Identifies a Novel <i>FOXP3</i> Mutation in a 2â€Generation Family With Inflammatory Bowel Disease. Journal of Pediatric Gastroenterology and Nutrition, 2014, 58, 561-568.	1.8	47
51	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
52	Synaptic, transcriptional and chromatin genes disrupted in autism. Nature, 2014, 515, 209-215.	27.8	2,254
53	A Comprehensive Search for Recombinogenic Motifs in the Human Genome. PLoS ONE, 2013, 8, e62920.	2.5	O
54	Multiplex Chromosomal Exome Sequencing Accelerates Identification of ENU-Induced Mutations in the Mouse. G3: Genes, Genomes, Genetics, 2012, 2, 143-150.	1.8	25

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55	Autosomal Recessive Causes Likely in Early-Onset Alzheimer Disease. Archives of Neurology, 2012, 69, 59.	4.5	193
56	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
57	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
58	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
59	Common NOD2 risk variants in African Americans with Crohn's disease are due exclusively to recent Caucasian admixture. Inflammatory Bowel Diseases, 2012, 18, 2357-2359.	1.9	18
60	Population Demographic History Can Cause the Appearance of Recombination Hotspots. American Journal of Human Genetics, 2012, 90, 774-783.	6.2	33
61	Targeted sequencing of the human X chromosome exome. Genomics, 2011, 98, 260-265.	2.9	22
62	Response to Graffelman: Tests of Hardy-Weinberg Equilibrium. American Journal of Human Genetics, 2010, 86, 818-819.	6.2	2
63	Microdeletions of 3q29 Confer High Risk for Schizophrenia. American Journal of Human Genetics, 2010, 87, 229-236.	6.2	215
64	SeqAnt: A web service to rapidly identify and annotate DNA sequence variations. BMC Bioinformatics, 2010, 11, 471.	2.6	38
65	Identification of novel <i>FMR1</i> variants by massively parallel sequencing in developmentally delayed males. American Journal of Medical Genetics, Part A, 2010, 152A, 2512-2520.	1.2	108
66	Array-Based FMR1 Sequencing and Deletion Analysis in Patients with a Fragile X Syndrome–Like Phenotype. PLoS ONE, 2010, 5, e9476.	2.5	26
67	Microarray oligonucleotide probe designer: a Web service. Open Access Bioinformatics, 2010, 2, 145.	0.9	5
68	To Pool, or Not to Pool?. Genetics, 2010, 186, 41-43.	2.9	86
69	Empirical Evaluation of Oligonucleotide Probe Selection for DNA Microarrays. PLoS ONE, 2010, 5, e9921.	2.5	14
70	Variation in the Lymphotoxin-α/Tumor Necrosis Factor Locus Modifies Risk of Erythema Nodosum in Sarcoidosis. Journal of Investigative Dermatology, 2009, 129, 1921-1926.	0.7	24
71	Validation and extension of an empirical Bayes method for SNP calling on Affymetrix microarrays. Genome Biology, 2008, 9, R63.	9.6	30
72	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

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73	Population Bottlenecks as a Potential Major Shaping Force of Human Genome Architecture. PLoS Genetics, 2007, 3, e119.	3.5	55
74	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
75	Investigating the role of p11 (S100A10) sequence variation in susceptibility to major depression. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2007, 144B, 1079-1082.	1.7	11
76	Microarray-based genomic selection for high-throughput resequencing. Nature Methods, 2007, 4, 907-909.	19.0	374
77	Genome-wide detection and characterization of positive selection in human populations. Nature, 2007, 449, 913-918.	27.8	1,788
78	A second generation human haplotype map of over 3.1 million SNPs. Nature, 2007, 449, 851-861.	27.8	4,137
79	An Oligonucleotide Microarray for High-Throughput Sequencing of the Mitochondrial Genome. Journal of Molecular Diagnostics, 2006, 8, 476-482.	2.8	65
80	Genomic alterations in cultured human embryonic stem cells. Nature Genetics, 2005, 37, 1099-1103.	21.4	592
81	A common sex-dependent mutation in a RET enhancer underlies Hirschsprung disease risk. Nature, 2005, 434, 857-863.	27.8	438
82	On the probability that a novel variant is a disease-causing mutation. Genome Research, 2005, 15, 960-966.	5.5	24
83	A Note on Exact Tests of Hardy-Weinberg Equilibrium. American Journal of Human Genetics, 2005, 76, 887-893.	6.2	1,232
84	Haplotype and Missing Data Inference in Nuclear Families. Genome Research, 2004, 14, 1624-1632.	5 . 5	42
85	Discrepancies in dbSNP confirmation rates and allele frequency distributions from varying genotyping error rates and patterns. Bioinformatics, 2004, 20, 1022-1032.	4.1	52
86	Tracking the Evolution of the SARS Coronavirus Using High-Throughput, High-Density Resequencing Arrays. Genome Research, 2004, 14, 398-405.	5 . 5	104
87	Exhaustive allelic transmission disequilibrium tests as a new approach to genome-wide association studies. Nature Genetics, 2004, 36, 1181-1188.	21.4	154
88	Microarray-based resequencing of multiple Bacillus anthracis isolates. Genome Biology, 2004, 6, R10.	9.6	64
89	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
90	Haplotype Inference in Random Population Samples. American Journal of Human Genetics, 2002, 71, 1129-1137.	6.2	176

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91	High-Throughput Variation Detection and Genotyping Using Microarrays. Genome Research, 2001, 11, 1913-1925.	5.5	258
92	Estimating Divergence Times in the Presence of an Overdispersed Molecular Clock. Molecular Biology and Evolution, 2000, 17, 1647-1660.	8.9	88
93	The Index of Dispersion of Molecular Evolution: Slow Fluctuations. Theoretical Population Biology, 2000, 57, 177-186.	1.1	8
94	PATTERNS OFGENETICVARIATION INMENDELIAN ANDCOMPLEXTRAITS. Annual Review of Genomics and Human Genetics, 2000, $1,387-407$.	6.2	78
95	Understanding the Overdispersed Molecular Clock. Genetics, 2000, 154, 1403-1417.	2.9	64
96	Classic Weinstein: Tetrad Analysis, Genetic Variation and Achiasmate Segregation in Drosophila and Humans. Genetics, 1999, 152, 1615-1629.	2.9	21
97	Clustered Mutations Have No Effect on the Overdispersed Molecular Clock: A Response to Huai and Woodruff. Genetics, 1998, 149, 463-464.	2.9	2
98	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