

Carlos D Bustamante

List of Publications by Year in descending order

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Version: 2024-02-01

90
papers

12,121
citations

53939

47
h-index

56606

87
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93
all docs

93
docs citations

93
times ranked

24399
citing authors

#	ARTICLE	IF	CITATIONS
1	REVEL: An Ensemble Method for Predicting the Pathogenicity of Rare Missense Variants. American Journal of Human Genetics, 2016, 99, 877-885.	2.6	1,555
2	Human Demographic History Impacts Genetic Risk Prediction across Diverse Populations. American Journal of Human Genetics, 2017, 100, 635-649.	2.6	1,120
3	Genomics for the world. Nature, 2011, 475, 163-165.	13.7	523
4	The genome of a Late Pleistocene human from a Clovis burial site in western Montana. Nature, 2014, 506, 225-229.	13.7	500
5	Genome Sequencing Highlights the Dynamic Early History of Dogs. PLoS Genetics, 2014, 10, e1004016.	1.5	481
6	Genomic evidence for the Pleistocene and recent population history of Native Americans. Science, 2015, 349, aab3884.	6.0	449
7	The genetics of Mexico recapitulates Native American substructure and affects biomedical traits. Science, 2014, 344, 1280-1285.	6.0	420
8	Genetic Control of Chromatin States in Humans Involves Local and Distal Chromosomal Interactions. Cell, 2015, 162, 1051-1065.	13.5	304
9	Pulling out the 1%: Whole-Genome Capture for the Targeted Enrichment of Ancient DNA Sequencing Libraries. American Journal of Human Genetics, 2013, 93, 852-864.	2.6	284
10	Punctuated bursts in human male demography inferred from 1,244 worldwide Y-chromosome sequences. Nature Genetics, 2016, 48, 593-599.	9.4	273
11	Spectrum and prevalence of genetic predisposition in medulloblastoma: a retrospective genetic study and prospective validation in a clinical trial cohort. Lancet Oncology, The, 2018, 19, 785-798.	5.1	268
12	Effect of predicted protein-truncating genetic variants on the human transcriptome. Science, 2015, 348, 666-669.	6.0	252
13	The ancestry and affiliations of Kennewick Man. Nature, 2015, 523, 455-458.	13.7	241
14	Estimating the mutation load in human genomes. Nature Reviews Genetics, 2015, 16, 333-343.	7.7	233
15	Distance from sub-Saharan Africa predicts mutational load in diverse human genomes. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, E440-9.	3.3	224
16	Comprehensive transcriptome analysis using synthetic long-read sequencing reveals molecular co-association of distant splicing events. Nature Biotechnology, 2015, 33, 736-742.	9.4	205
17	The African Turquoise Killifish Genome Provides Insights into Evolution and Genetic Architecture of Lifespan. Cell, 2015, 163, 1539-1554.	13.5	200
18	Genetic Recombination Is Targeted towards Gene Promoter Regions in Dogs. PLoS Genetics, 2013, 9, e1003984.	1.5	198

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19	Genomic Insights into the Ancestry and Demographic History of South America. PLoS Genetics, 2015, 11, e1005602.	1.5	198
20	Genetic structure in village dogs reveals a Central Asian domestication origin. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 13639-13644.	3.3	192
21	Dispersals and genetic adaptation of Bantu-speaking populations in Africa and North America. Science, 2017, 356, 543-546.	6.0	188
22	Reconstructing Native American Migrations from Whole-Genome and Whole-Exome Data. PLoS Genetics, 2013, 9, e1004023.	1.5	185
23	The Great Migration and African-American Genomic Diversity. PLoS Genetics, 2016, 12, e1006059.	1.5	166
24	Privacy Risks from Genomic Data-Sharing Beacons. American Journal of Human Genetics, 2015, 97, 631-646.	2.6	161
25	A continuum of admixture in the Western Hemisphere revealed by the African Diaspora genome. Nature Communications, 2016, 7, 12522.	5.8	136
26	An Unexpectedly Complex Architecture for Skin Pigmentation in Africans. Cell, 2017, 171, 1340-1353.e14.	13.5	134
27	Ancient genomes from North Africa evidence prehistoric migrations to the Maghreb from both the Levant and Europe. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 6774-6779.	3.3	131
28	Compact genome of the Antarctic midge is likely an adaptation to an extreme environment. Nature Communications, 2014, 5, 4611.	5.8	128
29	The phylogenetic and geographic structure of Y-chromosome haplogroup R1a. European Journal of Human Genetics, 2015, 23, 124-131.	1.4	122
30	Genetic ancestry influences asthma susceptibility and lung function among Latinos. Journal of Allergy and Clinical Immunology, 2015, 135, 228-235.	1.5	113
31	Genome-wide ancestry of 17th-century enslaved Africans from the Caribbean. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 3669-3673.	3.3	110
32	A genome-wide association study of bronchodilator response in Latinos implicates rare variants. Journal of Allergy and Clinical Immunology, 2014, 133, 370-378.e15.	1.5	105
33	Polygenic risk scores: a biased prediction?. Genome Medicine, 2018, 10, 100.	3.6	104
34	Genetic Mapping and Biochemical Basis of Yellow Feather Pigmentation in Budgerigars. Cell, 2017, 171, 427-439.e21.	13.5	101
35	Multidimensional structure-function relationships in human β -cardiac myosin from population-scale genetic variation. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 6701-6706.	3.3	98
36	The Time Scale of Recombination Rate Evolution in Great Apes. Molecular Biology and Evolution, 2016, 33, 928-945.	3.5	92

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37	An Efficient Multiple-Testing Adjustment for eQTL Studies that Accounts for Linkage Disequilibrium between Variants. <i>American Journal of Human Genetics</i> , 2016, 98, 216-224.	2.6	91
38	Patterns of genomic and phenomic diversity in wine and table grapes. <i>Horticulture Research</i> , 2017, 4, 17035.	2.9	87
39	Population Genomic Analysis of Ancient and Modern Genomes Yields New Insights into the Genetic Ancestry of the Tyrolean Iceman and the Genetic Structure of Europe. <i>PLoS Genetics</i> , 2014, 10, e1004353.	1.5	86
40	The Divergence of Neandertal and Modern Human Y Chromosomes. <i>American Journal of Human Genetics</i> , 2016, 98, 728-734.	2.6	81
41	Demographically-Based Evaluation of Genomic Regions under Selection in Domestic Dogs. <i>PLoS Genetics</i> , 2016, 12, e1005851.	1.5	77
42	Extreme selective sweeps independently targeted the X chromosomes of the great apes. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, 6413-6418.	3.3	75
43	Population genomic analyses of the chocolate tree, <i>Theobroma cacao</i> L., provide insights into its domestication process. <i>Communications Biology</i> , 2018, 1, 167.	2.0	73
44	Strategies for Enriching Variant Coverage in Candidate Disease Loci on a Multiethnic Genotyping Array. <i>PLoS ONE</i> , 2016, 11, e0167758.	1.1	72
45	Mechanisms Underlying Adaptation to Life in Hydrogen Sulfide-Rich Environments. <i>Molecular Biology and Evolution</i> , 2016, 33, 1419-1434.	3.5	69
46	Origins and genetic legacies of the Caribbean Taino. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, 2341-2346.	3.3	64
47	Genetic variant in folate homeostasis is associated with lower warfarin dose in African Americans. <i>Blood</i> , 2014, 124, 2298-2305.	0.6	57
48	Xrare: a machine learning method jointly modeling phenotypes and genetic evidence for rare disease diagnosis. <i>Genetics in Medicine</i> , 2019, 21, 2126-2134.	1.1	56
49	Inference of Gorilla Demographic and Selective History from Whole-Genome Sequence Data. <i>Molecular Biology and Evolution</i> , 2015, 32, 600-612.	3.5	55
50	Mitogenomes illuminate the origin and migration patterns of the indigenous people of the Canary Islands. <i>PLoS ONE</i> , 2019, 14, e0209125.	1.1	54
51	Genome-wide association study and admixture mapping reveal new loci associated with total IgE levels in Latinos. <i>Journal of Allergy and Clinical Immunology</i> , 2015, 135, 1502-1510.	1.5	52
52	Clinical Genetics Lacks Standard Definitions and Protocols for the Collection and Use of Diversity Measures. <i>American Journal of Human Genetics</i> , 2020, 107, 72-82.	2.6	52
53	Transcriptome Sequencing from Diverse Human Populations Reveals Differentiated Regulatory Architecture. <i>PLoS Genetics</i> , 2014, 10, e1004549.	1.5	49
54	Comparative performance of two whole-genome capture methodologies on ancient DNA Illumina libraries. <i>Methods in Ecology and Evolution</i> , 2015, 6, 725-734.	2.2	43

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55	Population History and Gene Divergence in Native Mexicans Inferred from 76 Human Exomes. <i>Molecular Biology and Evolution</i> , 2020, 37, 994-1006.	3.5	43
56	Toward Genetics-Driven Early Intervention in Dilated Cardiomyopathy. <i>Circulation: Cardiovascular Genetics</i> , 2017, 10, .	5.1	41
57	Genomic Evidence for Local Adaptation of Hunter-Gatherers to the African Rainforest. <i>Current Biology</i> , 2019, 29, 2926-2935.e4.	1.8	40
58	Discovery and functional characterization of a neomorphic PTEN mutation. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, 13976-13981.	3.3	38
59	Efficient analysis of large datasets and sex bias with ADMIXTURE. <i>BMC Bioinformatics</i> , 2016, 17, 218.	1.2	38
60	The Genome of the Self-Fertilizing Mangrove Rivulus Fish, <i>Kryptolebias marmoratus</i> : A Model for Studying Phenotypic Plasticity and Adaptations to Extreme Environments. <i>Genome Biology and Evolution</i> , 2016, 8, 2145-2154.	1.1	38
61	A research roadmap for next-generation sequencing informatics. <i>Science Translational Medicine</i> , 2016, 8, 335ps10.	5.8	37
62	A genome-wide association study identifies only two ancestry specific variants associated with spontaneous preterm birth. <i>Scientific Reports</i> , 2018, 8, 226.	1.6	37
63	Imputation-Aware Tag SNP Selection To Improve Power for Large-Scale, Multi-ethnic Association Studies. <i>G3: Genes, Genomes, Genetics</i> , 2018, 8, 3255-3267.	0.8	36
64	ClinGen Variant Curation Interface: a variant classification platform for the application of evidence criteria from ACMG/AMP guidelines. <i>Genome Medicine</i> , 2022, 14, 6.	3.6	34
65	An admixture mapping meta-analysis implicates genetic variation at 18q21 with asthma susceptibility in Latinos. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 143, 957-969.	1.5	33
66	Phylogenetic applications of whole Y-chromosome sequences and the Near Eastern origin of Ashkenazi Levites. <i>Nature Communications</i> , 2013, 4, 2928.	5.8	31
67	GBStools: A Statistical Method for Estimating Allelic Dropout in Reduced Representation Sequencing Data. <i>PLoS Genetics</i> , 2016, 12, e1005631.	1.5	30
68	Genetic variation drives seasonal onset of hibernation in the 13-lined ground squirrel. <i>Communications Biology</i> , 2019, 2, 478.	2.0	28
69	FasTag: Automatic text classification of unstructured medical narratives. <i>PLoS ONE</i> , 2020, 15, e0234647.	1.1	23
70	Gene expression imputation identifies candidate genes and susceptibility loci associated with cutaneous squamous cell carcinoma. <i>Nature Communications</i> , 2018, 9, 4264.	5.8	21
71	In-solution Y-chromosome capture-enrichment on ancient DNA libraries. <i>BMC Genomics</i> , 2018, 19, 608.	1.2	20
72	Whole-genome sequencing of Atacama skeleton shows novel mutations linked with dysplasia. <i>Genome Research</i> , 2018, 28, 423-431.	2.4	19

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73	Development of a small panel of SNPs to infer ancestry in Chileans that distinguishes Aymara and Mapuche components. <i>Biological Research</i> , 2020, 53, 15.	1.5	18
74	DeepTag: inferring diagnoses from veterinary clinical notes. <i>Npj Digital Medicine</i> , 2018, 1, 60.	5.7	17
75	Rapid evolution of a skin-lightening allele in southern African KhoeSan. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, 13324-13329.	3.3	17
76	Whole-genome sequencing reveals the extent of heterozygosity in a preferentially self-fertilizing hermaphroditic vertebrate. <i>Genome</i> , 2018, 61, 241-247.	0.9	15
77	Inexpensive and Highly Reproducible Cloud-Based Variant Calling of 2,535 Human Genomes. <i>PLoS ONE</i> , 2015, 10, e0129277.	1.1	14
78	A genetic counseling needs assessment of Mexico. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e668.	0.6	12
79	Structural Variation Detection by Proximity Ligation from Formalin-Fixed, Paraffin-Embedded Tumor Tissue. <i>Journal of Molecular Diagnostics</i> , 2019, 21, 375-383.	1.2	10
80	Clotting factor genes are associated with preeclampsia in high-altitude pregnant women in the Peruvian Andes. <i>American Journal of Human Genetics</i> , 2022, 109, 1117-1139.	2.6	10
81	PATH-SCAN: A REPORTING TOOL FOR IDENTIFYING CLINICALLY ACTIONABLE VARIANTS. , 2013, , .		9
82	The inference of sex-biased human demography from whole-genome data. <i>PLoS Genetics</i> , 2019, 15, e1008293.	1.5	7
83	IMPUTATION-BASED ASSESSMENT OF NEXT GENERATION RARE EXOME VARIANT ARRAYS. , 2013, , .		6
84	Using genotype array data to compare multi- and single-sample variant calls and improve variant call sets from deep coverage whole-genome sequencing data. <i>Bioinformatics</i> , 2016, 33, btw786.	1.8	3
85	Identification of a Novel Somatic Mutation Leading to Allele Dropout for EGFR L858R Genotyping in Non-Small Cell Lung Cancer. <i>Molecular Diagnosis and Therapy</i> , 2017, 21, 431-436.	1.6	2
86	Discovering prescription patterns in pediatric acute-onset neuropsychiatric syndrome patients. <i>Journal of Biomedical Informatics</i> , 2021, 113, 103664.	2.5	2
87	Bayesian model comparison for rare-variant association studies. <i>American Journal of Human Genetics</i> , 2021, 108, 2354-2367.	2.6	2
88	Beyond the reference genome. <i>Nature Biotechnology</i> , 2015, 33, 605-606.	9.4	1
89	Reply to Wang et al.: Sequencing datasets do not refute Central Asian domestication origin of dogs. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, E2556-E2557.	3.3	1
90	Data mining of digitized health records in a resource-constrained setting reveals that timely immunophenotyping is associated with improved breast cancer outcomes. <i>BMC Cancer</i> , 2018, 18, 933.	1.1	1