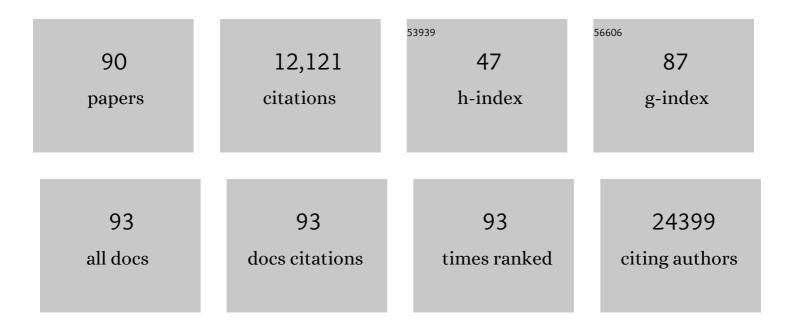
## Carlos D Bustamante

List of Publications by Year in descending order

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CAPLOS D RUSTAMANTE

| #  | Article  | IF  | CITATIONS |
|----|--|-----|-----------|
| 1  | ClinGen Variant Curation Interface: a variant classification platform for the application of evidence criteria from ACMG/AMP guidelines. Genome Medicine, 2022, 14, 6.             | 3.6 | 34        |
| 2  | Clotting factor genes are associated with preeclampsia in high-altitude pregnant women in the<br>Peruvian Andes. American Journal of Human Genetics, 2022, 109, 1117-1139.         | 2.6 | 10        |
| 3  | Discovering prescription patterns in pediatric acute-onset neuropsychiatric syndrome patients.<br>Journal of Biomedical Informatics, 2021, 113, 103664.                            | 2.5 | 2         |
| 4  | Bayesian model comparison for rare-variant association studies. American Journal of Human Genetics, 2021, 108, 2354-2367.  | 2.6 | 2         |
| 5  | Population History and Gene Divergence in Native Mexicans Inferred from 76 Human Exomes.<br>Molecular Biology and Evolution, 2020, 37, 994-1006.                                   | 3.5 | 43        |
| 6  | Clinical Genetics Lacks Standard Definitions and Protocols for the Collection and Use of Diversity<br>Measures. American Journal of Human Genetics, 2020, 107, 72-82.              | 2.6 | 52        |
| 7  | FasTag: Automatic text classification of unstructured medical narratives. PLoS ONE, 2020, 15, e0234647.  | 1.1 | 23        |
| 8  | Development of a small panel of SNPs to infer ancestry in Chileans that distinguishes Aymara and<br>Mapuche components. Biological Research, 2020, 53, 15.                         | 1.5 | 18        |
| 9  | The inference of sex-biased human demography from whole-genome data. PLoS Genetics, 2019, 15, e1008293.  | 1.5 | 7         |
| 10 | Genomic Evidence for Local Adaptation of Hunter-Gatherers to the African Rainforest. Current<br>Biology, 2019, 29, 2926-2935.e4.   | 1.8 | 40        |
| 11 | Xrare: a machine learning method jointly modeling phenotypes and genetic evidence for rare disease diagnosis. Genetics in Medicine, 2019, 21, 2126-2134.                           | 1.1 | 56        |
| 12 | A genetic counseling needs assessment of Mexico. Molecular Genetics & Genomic Medicine, 2019, 7,<br>e668.  | 0.6 | 12        |
| 13 | Mitogenomes illuminate the origin and migration patterns of the indigenous people of the Canary<br>Islands. PLoS ONE, 2019, 14, e0209125.  | 1.1 | 54        |
| 14 | Genetic variation drives seasonal onset of hibernation in the 13-lined ground squirrel.<br>Communications Biology, 2019, 2, 478.   | 2.0 | 28        |
| 15 | An admixture mapping meta-analysis implicates genetic variation at 18q21 with asthma susceptibility in<br>Latinos. Journal of Allergy and Clinical Immunology, 2019, 143, 957-969. | 1.5 | 33        |
| 16 | Structural Variation Detection by Proximity Ligation from Formalin-Fixed, Paraffin-Embedded Tumor<br>Tissue. Journal of Molecular Diagnostics, 2019, 21, 375-383.                  | 1.2 | 10        |
| 17 | Origins and genetic legacies of the Caribbean Taino. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 2341-2346.                        | 3.3 | 64        |
| 18 | A genome-wide association study identifies only two ancestry specific variants associated with spontaneous preterm birth. Scientific Reports, 2018, 8, 226.                        | 1.6 | 37        |

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|----|--|------|-----------|
| 19 | Whole-genome sequencing reveals the extent of heterozygosity in a preferentially self-fertilizing hermaphroditic vertebrate. Genome, 2018, 61, 241-247.  | 0.9  | 15        |
| 20 | Whole-genome sequencing of Atacama skeleton shows novel mutations linked with dysplasia. Genome Research, 2018, 28, 423-431.   | 2.4  | 19        |
| 21 | Imputation-Aware Tag SNP Selection To Improve Power for Large-Scale, Multi-ethnic Association Studies. G3: Genes, Genomes, Genetics, 2018, 8, 3255-3267.   | 0.8  | 36        |
| 22 | DeepTag: inferring diagnoses from veterinary clinical notes. Npj Digital Medicine, 2018, 1, 60.  | 5.7  | 17        |
| 23 | Polygenic risk scores: a biased prediction?. Genome Medicine, 2018, 10, 100.   | 3.6  | 104       |
| 24 | Rapid evolution of a skin-lightening allele in southern African KhoeSan. Proceedings of the National<br>Academy of Sciences of the United States of America, 2018, 115, 13324-13329.   | 3.3  | 17        |
| 25 | Data mining of digitized health records in a resource-constrained setting reveals that timely immunophenotyping is associated with improved breast cancer outcomes. BMC Cancer, 2018, 18, 933.                                 | 1.1  | 1         |
| 26 | Population genomic analyses of the chocolate tree, Theobroma cacao L., provide insights into its domestication process. Communications Biology, 2018, 1, 167.  | 2.0  | 73        |
| 27 | Gene expression imputation identifies candidate genes and susceptibility loci associated with cutaneous squamous cell carcinoma. Nature Communications, 2018, 9, 4264.   | 5.8  | 21        |
| 28 | 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       |
| 29 | In-solution Y-chromosome capture-enrichment on ancient DNA libraries. BMC Genomics, 2018, 19, 608.   | 1.2  | 20        |
| 30 | Ancient genomes from North Africa evidence prehistoric migrations to the Maghreb from both the<br>Levant and Europe. Proceedings of the National Academy of Sciences of the United States of America,<br>2018, 115, 6774-6779. | 3.3  | 131       |
| 31 | Dispersals and genetic adaptation of Bantu-speaking populations in Africa and North America. Science, 2017, 356, 543-546.  | 6.0  | 188       |
| 32 | Identification of a Novel Somatic Mutation Leading to Allele Dropout for EGFR L858R Genotyping in<br>Non-Small Cell Lung Cancer. Molecular Diagnosis and Therapy, 2017, 21, 431-436.   | 1.6  | 2         |
| 33 | Human Demographic History Impacts Genetic Risk Prediction across Diverse Populations. American<br>Journal of Human Genetics, 2017, 100, 635-649.   | 2.6  | 1,120     |
| 34 | Genetic Mapping and Biochemical Basis of Yellow Feather Pigmentation in Budgerigars. Cell, 2017, 171, 427-439.e21.   | 13.5 | 101       |
| 35 | Patterns of genomic and phenomic diversity in wine and table grapes. Horticulture Research, 2017, 4, 17035.  | 2.9  | 87        |
| 36 | Toward Genetics-Driven Early Intervention in Dilated Cardiomyopathy. Circulation: Cardiovascular<br>Genetics, 2017, 10, .  | 5.1  | 41        |

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|----|--|------|-----------|
| 37 | An Unexpectedly Complex Architecture for Skin Pigmentation in Africans. Cell, 2017, 171, 1340-1353.e14.  | 13.5 | 134       |
| 38 | Efficient analysis of large datasets and sex bias with ADMIXTURE. BMC Bioinformatics, 2016, 17, 218.   | 1.2  | 38        |
| 39 | GBStools: A Statistical Method for Estimating Allelic Dropout in Reduced Representation Sequencing<br>Data. PLoS Genetics, 2016, 12, e1005631.   | 1.5  | 30        |
| 40 | The Great Migration and African-American Genomic Diversity. PLoS Genetics, 2016, 12, e1006059.   | 1.5  | 166       |
| 41 | Using genotype array data to compare multi- and single-sample variant calls and improve variant call sets from deep coverage whole-genome sequencing data. Bioinformatics, 2016, 33, btw786.   | 1.8  | 3         |
| 42 | Mechanisms Underlying Adaptation to Life in Hydrogen Sulfide–Rich Environments. Molecular<br>Biology and Evolution, 2016, 33, 1419-1434.   | 3.5  | 69        |
| 43 | The Divergence of Neandertal and Modern Human Y Chromosomes. American Journal of Human<br>Genetics, 2016, 98, 728-734.   | 2.6  | 81        |
| 44 | Reply to Wang et al.: Sequencing datasets do not refute Central Asian domestication origin of dogs.<br>Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, E2556-E2557.                        | 3.3  | 1         |
| 45 | Punctuated bursts in human male demography inferred from 1,244 worldwide Y-chromosome sequences. Nature Genetics, 2016, 48, 593-599.   | 9.4  | 273       |
| 46 | REVEL: An Ensemble Method for Predicting the Pathogenicity of Rare Missense Variants. American<br>Journal of Human Genetics, 2016, 99, 877-885.  | 2.6  | 1,555     |
| 47 | The Genome of the Self-Fertilizing Mangrove Rivulus Fish, <i>Kryptolebias marmoratus</i> : A Model<br>for Studying Phenotypic Plasticity and Adaptations to Extreme Environments. Genome Biology and<br>Evolution, 2016, 8, 2145-2154. | 1.1  | 38        |
| 48 | A continuum of admixture in the Western Hemisphere revealed by the African Diaspora genome. Nature<br>Communications, 2016, 7, 12522.  | 5.8  | 136       |
| 49 | A research roadmap for next-generation sequencing informatics. Science Translational Medicine, 2016, 8, 335ps10.   | 5.8  | 37        |
| 50 | 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        |
| 51 | An Efficient Multiple-Testing Adjustment for eQTL Studies that Accounts for Linkage Disequilibrium between Variants. American Journal of Human Genetics, 2016, 98, 216-224.  | 2.6  | 91        |
| 52 | Distance from sub-Saharan Africa predicts mutational load in diverse human genomes. Proceedings of the United States of America, 2016, 113, E440-9.  | 3.3  | 224       |
| 53 | The Time Scale of Recombination Rate Evolution in Great Apes. Molecular Biology and Evolution, 2016, 33, 928-945.  | 3.5  | 92        |
| 54 | Demographically-Based Evaluation of Genomic Regions under Selection in Domestic Dogs. PLoS<br>Genetics, 2016, 12, e1005851.  | 1.5  | 77        |

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|----|---|------|-----------|
| 55 | Strategies for Enriching Variant Coverage in Candidate Disease Loci on a Multiethnic Genotyping<br>Array. PLoS ONE, 2016, 11, e0167758.   | 1.1  | 72        |
| 56 | Privacy Risks from Genomic Data-Sharing Beacons. American Journal of Human Genetics, 2015, 97, 631-646.   | 2.6  | 161       |
| 57 | Comparative performance of two wholeâ€genome capture methodologies on ancient <scp>DNA</scp><br>Illumina libraries. Methods in Ecology and Evolution, 2015, 6, 725-734.                     | 2.2  | 43        |
| 58 | Genomic Insights into the Ancestry and Demographic History of South America. PLoS Genetics, 2015, 11, e1005602.   | 1.5  | 198       |
| 59 | Extreme selective sweeps independently targeted the X chromosomes of the great apes. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 6413-6418. | 3.3  | 75        |
| 60 | Beyond the reference genome. Nature Biotechnology, 2015, 33, 605-606.   | 9.4  | 1         |
| 61 | Inference of Gorilla Demographic and Selective History from Whole-Genome Sequence Data.<br>Molecular Biology and Evolution, 2015, 32, 600-612.  | 3.5  | 55        |
| 62 | Genome-wide association study and admixture mapping reveal new loci associated with total IgE levels<br>in Latinos. Journal of Allergy and Clinical Immunology, 2015, 135, 1502-1510.       | 1.5  | 52        |
| 63 | 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       |
| 64 | The African Turquoise Killifish Genome Provides Insights into Evolution and Genetic Architecture of<br>Lifespan. Cell, 2015, 163, 1539-1554.  | 13.5 | 200       |
| 65 | Genetic ancestry influences asthma susceptibility and lung function among Latinos. Journal of<br>Allergy and Clinical Immunology, 2015, 135, 228-235.                                       | 1.5  | 113       |
| 66 | Genomic evidence for the Pleistocene and recent population history of Native Americans. Science, 2015, 349, aab3884.  | 6.0  | 449       |
| 67 | The ancestry and affiliations of Kennewick Man. Nature, 2015, 523, 455-458.   | 13.7 | 241       |
| 68 | 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       |
| 69 | Estimating the mutation load in human genomes. Nature Reviews Genetics, 2015, 16, 333-343.  | 7.7  | 233       |
| 70 | Effect of predicted protein-truncating genetic variants on the human transcriptome. Science, 2015, 348, 666-669.  | 6.0  | 252       |
| 71 | Discovery and functional characterization of a neomorphic PTEN mutation. Proceedings of the<br>National Academy of Sciences of the United States of America, 2015, 112, 13976-13981.        | 3.3  | 38        |
| 72 | Genetic structure in village dogs reveals a Central Asian domestication origin. Proceedings of the<br>National Academy of Sciences of the United States of America, 2015, 112, 13639-13644. | 3.3  | 192       |

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|----|---|------|-----------|
| 73 | Genetic Control of Chromatin States in Humans Involves Local and Distal Chromosomal Interactions.<br>Cell, 2015, 162, 1051-1065.  | 13.5 | 304       |
| 74 | The phylogenetic and geographic structure of Y-chromosome haplogroup R1a. European Journal of<br>Human Genetics, 2015, 23, 124-131.   | 1.4  | 122       |
| 75 | Inexpensive and Highly Reproducible Cloud-Based Variant Calling of 2,535 Human Genomes. PLoS ONE, 2015, 10, e0129277.   | 1.1  | 14        |
| 76 | Transcriptome Sequencing from Diverse Human Populations Reveals Differentiated Regulatory<br>Architecture. PLoS Genetics, 2014, 10, e1004549.   | 1.5  | 49        |
| 77 | Genome Sequencing Highlights the Dynamic Early History of Dogs. PLoS Genetics, 2014, 10, e1004016.  | 1.5  | 481       |
| 78 | Population Genomic Analysis of Ancient and Modern Genomes Yields New Insights into the Genetic<br>Ancestry of the Tyrolean Iceman and the Genetic Structure of Europe. PLoS Genetics, 2014, 10, e1004353. | 1.5  | 86        |
| 79 | A genome-wide association study of bronchodilator response in Latinos implicates rare variants.<br>Journal of Allergy and Clinical Immunology, 2014, 133, 370-378.e15.                                    | 1.5  | 105       |
| 80 | The genome of a Late Pleistocene human from a Clovis burial site in western Montana. Nature, 2014,<br>506, 225-229.   | 13.7 | 500       |
| 81 | Compact genome of the Antarctic midge is likely an adaptation to an extreme environment. Nature<br>Communications, 2014, 5, 4611.   | 5.8  | 128       |
| 82 | The genetics of Mexico recapitulates Native American substructure and affects biomedical traits.<br>Science, 2014, 344, 1280-1285.  | 6.0  | 420       |
| 83 | Genetic variant in folate homeostasis is associated with lower warfarin dose in African Americans.<br>Blood, 2014, 124, 2298-2305.  | 0.6  | 57        |
| 84 | Pulling out the 1%: Whole-Genome Capture for the Targeted Enrichment of Ancient DNA Sequencing<br>Libraries. American Journal of Human Genetics, 2013, 93, 852-864.                                       | 2.6  | 284       |
| 85 | Phylogenetic applications of whole Y-chromosome sequences and the Near Eastern origin of Ashkenazi Levites. Nature Communications, 2013, 4, 2928.   | 5.8  | 31        |
| 86 | Genetic Recombination Is Targeted towards Gene Promoter Regions in Dogs. PLoS Genetics, 2013, 9, e1003984.  | 1.5  | 198       |
| 87 | Reconstructing Native American Migrations from Whole-Genome and Whole-Exome Data. PLoS Genetics, 2013, 9, e1004023.   | 1.5  | 185       |
| 88 | PATH-SCAN: A REPORTING TOOL FOR IDENTIFYING CLINICALLY ACTIONABLE VARIANTS. , 2013, , .   |      | 9         |
| 89 | IMPUTATION-BASED ASSESSMENT OF NEXT GENERATION RARE EXOME VARIANT ARRAYS. , 2013, , .   |      | 6         |
|    |   |      |           |