JONATHAN K. PRITCHARD

Bing Professor of Population Studies
Departments of Biology and Genetics
Stanford University

Web: http://pritchardlab.stanford.edu
Email: pritch@stanford.edu
CV version: May 2020

Employment History

2020–Present	Bing Professor of Population Studies
2013–Present	Professor, Departments of Genetics and Biology, Stanford University
2008–2019	Investigator, Howard Hughes Medical Institute
2006–2013	Professor, Department of Human Genetics, The University of Chicago
2001–2005	Assistant Professor, Department of Human Genetics, The University of Chicago

Education

1998–2001	Postdoctoral Fellow, Statistics University of Oxford Advised by Peter Donnelly
1994–1998	Ph.D., Biology Stanford University Advised by Marcus Feldman
1989–1994	B.Sc., Biology and Mathematics Pennsylvania State University

Awards and honors

2019	Fabio Frassetto International Prize in Physical Anthropology from the Lincean Academy of Italy
2016	Review of Pritchard et al. (2000) in the Classic Papers series for the centennial year of *Genetics* by John Novembre (PMID 27729489). P. et al. has now been cited >26,000 times (G Scholar)
2015	Kistler Prize in Population Genetics and Society
2013	Selected as a Fellow of the American Academy of Arts and Sciences
2013	Edward Novitski Prize from the Genetics Society of America; this award recognizes "creativity and intellectual ingenuity in the solution of significant problems in genetics"
2013	Outstanding Alumni Award, Eberly College of Science, Penn State University
2013	*Best postdoc talk* award to my postdoc S. Gopalakrishnan; Am. Soc. Hum Genet. meeting
2008	Selected as an investigator of the Howard Hughes Medical Institute
2007	*Best postdoc talk* award to my postdoc Graham Coop; Am. Soc. Hum. Genet. meeting.
2006	*Best Ph.D thesis award* to my student Ben Voight (for U of C Biological Sciences Division)
2006	New York Times front page news article on Voight et al paper on natural selection
2004	Selected Packard Fellow
2004	Selected Alfred P. Sloan Fellow

2003	Lancet's *Paper of the Year* (for any biomedical journal) was awarded to the paper of Rosenberg, Pritchard et al. (Science 2002) on human population structure (editorial in Lancet 362:2101-2103).
2002	Mitchell Prize from the American Statistical Association and the International Society of Bayesian Analysis, presented annually for "an outstanding paper describing how a Bayesian analysis has solved an important applied problem."
1994-1998	Howard Hughes Medical Institute Predoctoral Fellowship
1994	McCoy Award, given to the Penn State Outstanding Scholar-Athlete
1992	Stecker Award, given to a Penn State undergraduate in Mathematics
1989-1993	Penn State Braddock Scholarship and Academic Excellence Scholarship

Selected Professional Activities

2018–present	SAB/advisor: Calico, Precise.ly, Inari
2017–present	Co-Director: Stanford Center for Computational, Evolutionary and Human Genomics
2017–present	External Advisory Board to Wellcome Trust Sanger Institute
2017	Lead organizing cmte for "Human Evolution: fossils, archaeology, and genomics" meeting, Hinxton, UK
2016–2020	NHGRI (NIH) Council Member
2016–2018	Lead organizing committee for "Biology of Genomes" meeting, Cold Spring Harbor
2015–present	Science Magazine Board of Reviewing Editors
2014–present	Frequent guest editor for PLOS Genetics
2013	Scientific Advisory Board, Cellular Genomics Program, Wellcome Trust Sanger Institute
2012–present	Editorial Board: Current Biology
2008–2012	Associate Editor: Molecular Biology and Evolution
2006–2016	23andMe, SAB
2006–2014	Associate Editor: PLOS Genetics

Research Support [as PI]

2016-2024	RO1 HG008140: "Integration of functional data and GWAS to elucidate genetic basis of diseases"
2017-2020	UO1 HG009431: "Decoding the regulatory architecture of the human genome across cell types, individuals and disease."

Previous grants/fellowships:

2008-2019	Investigator, Howard Hughes Medical Institute.
2014-2017	RO1 ES025009: "Computational methods for modeling lineage-specific gene regulation." Administrative supplement for 2016-2017.
2012-2016	U01 HG007036: "Genetic and epigenetic controls of gene regulation." Includes administrative supplement for 2015-2016.
2010-2013	RO1 MH090951: "Statistical analysis of gene expression quantitative trait loci (eQTL)." Includes administrative supplement for 2012-2013.
2008-2016	RO1 MH084703: "Analysis and interpretation of DNA sequence data in association studies."
2003-2008	RO1 HG002772: "Linkage Disequilibrium Methods for Complex Trait Mapping."
2004–2009	Packard Foundation Fellowship: "Population genetics of genomic rearrangements."
2004-2006	Sloan Foundation Fellowship
1999-2003	Burroughs-Wellcome Fund Hitchings-Elion Postdoc-Faculty Award

	"Population structure and linkage disequilibrium in association mapping."
1998	NIH (NRSA) 3-year Postdoctoral Award (replaced by BWF award)
	"The impact of population history on association mapping."
1994-1998	Howard Hughes Medical Institute Predoctoral Fellowship
1994	NSF Predoctoral Fellowship (declined)
1989-1993	Penn State Braddock Scholarship (four-year full-tuition award)
1989-1993	Penn State Academic Excellence Scholarship

Graduate Student/Postdoc Mentoring:

2020–Present	Daphna Rothschild (postdoc; joint with Maria Barna)
2019–Present	Hakhamanesh Mostafavi (postdoc)
2019–Present	Clemens Weiss (postdoc)
2019–Present	Jeff Spence (postdoc)
2019–Present	Sahin Naqvi (postdoc; joint with Joanna Wysocka)
2019–Present	Shaila Musharoff (postdoc)
2018–Present	Yuval Simons (postdoc)
2018–Present	Jake Freimer (postdoc; joint with Alex Marson)
2019–Present	Alyssa Fortier (PhD student, Biology)
2019–Present	Roshni Patel (PhD student, Genetics)
2018–Present	Adele Xu (MSTP; primary lab Maria Barna)
2017–present	Hannah Moots (PhD student, Archaeology)
2017–present	Margaret Antonio (PhD student, BMI)
2017–present	Nasa Sinnott-Armstrong (PhD student, Genetics)

Previous Postdocs:

2018–2019	Hanna Ollila (postdoc); now Group Leader, University of Helsinki
2016–2020	Harold Pimentel (postdoc); now Assistant Professor, UCLA
2015–2020	Ziyue Gao (postdoc); now Assistant Professor, Univ Pennsylvania
2015–2018	David Knowles (postdoc); joint w/ Sylvia Plevritis; now asst prof, NY Genome Center/ Columbia
2014–2018	Eilon Sharon (postdoc); joint with Hunter Fraser; now staff scientist, Insitro
2012–2018	Yair Field (postdoc; now staff scientist, Illumina)
2015–2017	Kelley Harris (postdoc; now assistant professor, U Washington)
2012–2017	Xun Lan (postdoc; now assistant professor, Tsinghua University)
2014–2017	Yang Li (postdoc; now assistant professor, U Chicago)
2014–2017	Anand Bhaskar (postdoc); now data scientist, Facebook
2011–2016	Anil Raj (postdoc); now staff scientist, Calico
2014–2016	David Golan (postdoc); cofounder of Viz.ai
2014–2015	Audrey Fu (visiting postdoc); now assistant professor, Univ of Idaho
2014–2015	Towfique Raj (visiting postdoc); now assistant professor, Mt Sinai
2014–2015	Kyle Gaulton (visiting postdoc); now assistant professor, UCSD
2010–2015	Graham McVicker (postdoc); now assistant professor, Salk Institute
2013–2014	Alexis Battle (postdoc); now associate professor, Johns Hopkins
2011–2014	Stoyan Georgiev (postdoc); now data scientist, Google
2011–2013	Zia Khan (postdoc; joint with Y Gilad); assistant professor, University of Maryland; now Genentech
2011–2013	Shyam Gopalakrishnan (postdoc; joint with Abe Palmer); now Assoc Prof, U Copenhagen
2012–2013	Allegra Petti (postdoc; joint with Y Gilad); now staff scientist, Wash U St Louis
2009–2012	Roger Pique-Regi (postdoc); now associate professor, Wayne State, MI

2008–2011 Daniel Gaffney (postdoc); G.L. at Sanger Institute; now head of Cellular Genetics, Genomics PLC
2009–2011 Pall Melsted (postdoc); now assistant professor, University of Iceland
2009–2011 Ying Wang (postdoc); now junior faculty, Beijing Institute of Genomics
2008–2009 Jordana Bell (postdoc); now Senior Lecturer, Kings College London
2006–2007 Jean-Baptiste Veyrieras (postdoc); now group leader at bioMerieux
2004–2008 Graham Coop (postdoc); now professor, U. of California, Davis
2001–2005 Sebastian Zöllner (postdoc); now professor, Biostatistics, U. of Michigan
2002–2004 Giovanni Montana (postdoc); now professor of Biostatistics, Imperial College London
2002–2003 Jeffrey Wall (postdoc); now professor of Epidemiology and Biostats, UCSF

Previous PhD Students:

2015–2019 Jessica Ribado (PhD student, Genetics; joint w/ Ami Bhatt; now PD at Centers for Disease Modeling)
2015–2019 Evan Boyle (PhD student, Genetics; joint with Will Greenleaf; now postdoc with Gene Yeo, UCSD)
2014–2019 Diego Calderon (PhD student, BMI; now postdoc with Trapnell/Shendure at U Washington)
2014–2019 Emily Glassberg (PhD student, Biology; now at Boston Consulting Group)
2014–2018 Arbel Harpak (PhD student, Biology; postdoc with Molly Przeworski, Columbia)
2014–2018 Natalie Telis (PhD student, BMI; staff scientist at Ancestry)
2011–2015 Bryce van de Geijn (PhD student, Chicago GGSB); postdoc with Alkes Price, Harvard SPH
2014–2015 Cristina Pop (PhD, CS with Daphne Koller; mentored by me 2014-5); data scientist at Google
2011–2014 Paul Grabowski (PhD student, Ecology and Evolution; joint with J. Borevitz); postdoc at USDA
2012–2013 Carolyn Jumper (PhD student, Human Genetics); switched to Cox lab due to Stanford move
2012–2013 Michael Turchin (PhD student, Human Genetics); switched to Stephens lab due to Stanford move
2008–2012 Jack Degner (PhD, GGSB); postdoc with Eileen Furlong, EMBL; now Senior Scientist at AbbVie
2007–2011 Joseph Pickrell (PhD, Human Genetics); postdoc with David Reich, Harvard;
asst prof NY Genome Center/Columbia Univ; now founder/CEO of GenCove
2004–2008 Sridhar Kudaravalli (PhD, Human Genetics, 2008)
2004–2008 Su Yeon Kim (PhD, Statistics, 2008; joint with P. McCullagh)
postdoc with Rasmus Nielsen, Terry Speed at UC Berkeley; now Bioinformatician Veracyte
2004–2006 Daniel Davison (PhD, Comm Ev. Biol., 2006; joint with S. Hackett)
postdoc in Statistics with Peter Donnelly, U. of Oxford; now at Counsyl
2003–2007 Donald Conrad (PhD, Human Genetics, 2008)
postdoc with Matt Hurles, Sanger Institute; now assoc. prof. Wash U.
2002–2006 Benjamin Voight (PhD, Human Genetics, 2006; joint with N. Cox)
postdoc with Mark Daly, Broad Institute; now assoc. prof., Univ of Pennsylvania

Selected other lab members:

2001–2007 Ben Lehmann (programmer); now PhD student in astrophysics, UCSC
2011-2012 Andy Dahl (Honors Thesis, Statistics); DPhil student, Oxford; PD UCSF; now asst Prof U Chicago
2006–2008 Melissa Hubisz (M.S., Human Genetics); PhD student, Cornell; staff scientist Cornell
2001–2007 Xiaoquan William Wen (programmer); now Associate Professor Biostatistics, Michigan

Publications

[1] N Sinnott-Armstrong, S Naqvi, MA Rivas, and JK Pritchard. GWAS of three molecular traits highlights core genes and pathways alongside a highly polygenic background. *BioRxiv*, 2020.

[2] S Catania, PA Dumesic, H Pimentel, A Nasif, CI Stoddard, JE Burke, JK Diedrich, S Cook, T Shea, E Geinger, Lintner R, Yates JR 3rd, Hajkova P, GJ Narlikar, CA Cuomo, JK Pritchard, and HD. Madhani. Evolutionary persistence of DNA methylation for millions of years after ancient loss of a de novo methyltransferase. *Cell*, 2020.

[3] H Mostafavi, A Harpak, I Agarwal, D Conley, JK Pritchard, and M Przeworski. Variable prediction accuracy of polygenic scores within an ancestry group. *Elife*, 9:e48376, 2020.

[4] N Sinnott-Armstrong, Y Tanigawa, D Amar, NJ Mars, M Aguirre, GR Venkataraman, M Wainberg, HM Ollila, JP Pirruccello, J Qian, [8 others], JK Pritchard, MJ Daly, and MA Rivas. Genetics of 38 blood and urine biomarkers in the UK Biobank. *BioRxiv*, page 660506, 2019.

[5] AE Trevino, N Sinnott-Armstrong, J Andersen, S-J Yoon, N Huber, JK Pritchard, HY Chang, WJ Greenleaf, and SP Pasca. Chromatin accessibility dynamics in a model of human forebrain development. *Science*, 367(6476), 2020.

[6] ML Antonio, Z Gao, HM Moots, M Lucci, F Candilio, S Sawyer, V Oberreiter, D Calderon, K Devitofranceschi, RC Aikens, S Aneli, F Bartoli, A Bedini, O Cheronet, DJ Cotter, DM Fernandes, G Gasperetti, R Grifoni, A Guidi, F La Pastina, E Loreti, D Manacorda, G Matullo, S Morretta, A Nava, NV Fiocchi, F Nomi, C Pavolini, M Pentiricci, P Pergola, M Piranomonte, R Schmidt, G Spinola, A Sperduti, M Rubini, L Bondioli, A Coppa, R Pinhasi, and JK. Pritchard. Ancient Rome: A genetic crossroads of Europe and the Mediterranean. *Science*, 366(6466):708–714, 2019.

[7] D Calderon, MLT Nguyen, A Mezger, A Kathiria, F Müller, V Nguyen, N Lescano, B Wu, J Trombetta, JV Ribado, DA Knowles, Z Gao, F Blaeschke, AV Parent, TD Burt, MS Anderson, LA Criswell, WJ Greenleaf, A Marson, and JK. Pritchard. Landscape of stimulation-responsive chromatin across diverse human immune cells. *Nature Genetics*, pages 1–12, 2019.

[8] N Telis, EC Glassberg, JK Pritchard, and C Gunter. Public discussion affects question asking at academic conferences. *The American Journal of Human Genetics*, 2019.

[9] X Liu, YI Li, and JK Pritchard. Trans effects on gene expression can drive omnigenic inheritance. *Cell*, 177(4):1022–1034, 2019.

[10] MC Stahlschmidt, TC Collin, DM Fernandes, G Bar-Oz, A Belfer-Cohen, Z Gao, N Jakeli, Z Matskevich, T Meshveliani, JK Pritchard, F McDermott, and R Pinhasi. Ancient mammalian and plant DNA from late Quaternary stalagmite layers at Solkota Cave, Georgia. *Scientific Reports*, 9(1):6628, 2019.

[11] JJ Berg, A Harpak, N Sinnott-Armstrong, Anja M Joergensen, H Mostafavi, Y Field, EA Boyle, X Zhang, F Racimo, JK Pritchard, and Coop G. Reduced signal for polygenic adaptation of height in UK Biobank. *ELife*, 8:e39725, 2019.

[12] NA Rosenberg, MD Edge, JK Pritchard, and MW Feldman. Interpreting polygenic scores, polygenic adaptation, and human phenotypic differences. *Evolution, medicine, and public health*, 2019(1):26–34, 2019.

[13] EC Glassberg, Z Gao, A Harpak, X Lan, and JK Pritchard. Evidence for weak selective constraint on human gene expression. *Genetics*, 211(2):757–772, 2019.

[14] EA Boyle, JK Pritchard, and WJ Greenleaf. High-resolution mapping of cancer cell networks using co-functional interactions. *Molecular Systems Biology*, 14(12), 2018.

[15] H Ollila, E Sharon, L Lin, N Sinnott-Armstrong, A Ambati, RP Hillary, O Jolanki, J Faraco, M Einen, G Luo, et al. Narcolepsy risk loci are enriched in immune cells and suggest autoimmune modulation of the t cell receptor repertoire. *bioRxiv*, page 373555, 2018.

[16] P Desai, N Telis, B Lehmann, K Bettinger, JK Pritchard, and S Datta. Scireader*: A cloud-based recommender system for biomedical literature. *bioRxiv*, page 333922, 2018.

[17] E Sharon, S-AA Chen, NM Khosla, JD Smith, JK Pritchard, and HB Fraser. Functional genetic variants revealed by massively parallel precise genome editing. *Cell*, 175(2):544–557, 2018.

[18] SH Wang, CJ Hsiao, Z Khan, and JK Pritchard. Post-translational buffering leads to convergent protein expression levels between primates. *Genome Biology*, 19(1):83, 2018.

[19] DA Knowles, CK Burrows, JD Blischak, KM Patterson, DJ Serie, N Norton, C Ober, JK Pritchard, and Y Gilad. Determining the genetic basis of anthracycline-cardiotoxicity by molecular response QTL mapping in induced cardiomyocytes. *eLife*, 7, 2018.

[20] R Yamamoto, AC Wilkinson, J Ooehara, X Lan, C-Y Lai, Y Nakauchi, JK Pritchard, and H Nakauchi. Large-scale clonal analysis resolves aging of the mouse hematopoietic stem cell compartment. *Cell Stem Cell*, 22(4):600–607, 2018.

[21] D Sparvoli, E Richardson, H Osakada, X Lan, M Iwamoto, GR Bowman, C Kontur, WA Bourland, DH Lynn, JK Pritchard, T Haraguchi, JB Dacks, and Turkewitz AP. Remodeling the Specificity of an Endosomal CORVET Tether Underlies Formation of Regulated Secretory Vesicles in the Ciliate Tetrahymena thermophila. *Current Biology*, 28(5):697–710, 2018.

[22] YI Li, DA Knowles, J Humphrey, AN Barbeira, SP Dickinson, HK Im, and JK Pritchard. Annotation-free quantification of RNA splicing using LeafCutter. *Nature Genetics*, 50(1):151, 2018.

[23] NE Banovich, YI Li, A Raj, MC Ward, P Greenside, D Calderon, PY Tung, JE Burnett, M Myrthil, SM Thomas, CK Burrows, I Gallego Romero, BJ Pavlovic, A Kundaje, JK Pritchard, and Y Gilad. Impact of regulatory variation across human iPSCs and differentiated cells. *Genome Research*, 28(1):122–131, 2018.

[24] A Harpak, X Lan, Z Gao, and JK Pritchard. Frequent non-allelic gene conversion on the human lineage and its effect on the divergence of gene duplicates. *Proceedings of the National Academy of Sciences*, 114(48):12779–12784, 2017.

[25] D Calderon, A Bhaskar, DA Knowles, D Golan, T Raj, A Fu, and JK Pritchard. Inferring relevant cell types for complex traits using single-cell gene expression. *The American Journal of Human Genetics*, 101(5):686–699, 2017.

[26] E Sharon, H Shi, S Kharbanda, W Koh, LR Martin, KK Khush, H Valantine, JK Pritchard, and I De Vlaminck. Quantification of transplant-derived circulating cell-free DNA in absence of a donor genotype. *PLoS Computational Biology*, 13(8):e1005629, 2017.

[27] EA Boyle, YI Li, and JK Pritchard. The omnigenic model: Response from the authors. *Journal of Psychiatry and Brain Science*, 2(5):S8, 2017.

[28] EA Boyle, YI Li, and JK Pritchard. An expanded view of complex traits: From polygenic to omnigenic. *Cell*, 169(7):1177–1186, 2017.

[29] K Harris and JK Pritchard. Rapid evolution of the human mutation spectrum. *eLife*, 6:e24284, 2017.

[30] R Nielsen, JM Akey, M Jakobsson, JK Pritchard, S Tishkoff, and E Willerslev. Tracing the peopling of the world through genomics. *Nature*, 541(7637):302–310, 2017.

[31] PY Tung, JD Blischak, CJ Hsiao, DA Knowles, JE Burnett, JK Pritchard, and Y Gilad. Batch effects and the effective design of single-cell gene expression studies. *Scientific Reports*, 7, 2017.

[32] A Harpak, A Bhaskar, and JK Pritchard. Mutation rate variation is a primary determinant of the distribution of allele frequencies in humans. *PLoS Genetics*, 12(12):e1006489, 2016.

[33] N Telis, BV Lehmann, MW Feldman, and JK Pritchard. A bibliometric history of the journal Genetics. *Genetics*, 204(4):1337–1342, 2016.

[34] Y Field, EA Boyle, N Telis, Z Gao, KJ Gaulton, D Golan, L Yengo, G Rocheleau, P Froguel, MI McCarthy, and JK Pritchard. Detection of human adaptation during the past 2,000 years. *Science*, 354:760–764, 2016.

[35] MR Corces, JD Buenrostro, B Wu, PG Greenside, SM Chan, JL Koenig, MP Snyder, JK Pritchard, A Kundaje, WJ Greenleaf, R Majeti, and Chang HY. Lineage-specific and single-cell chromatin accessibility charts human hematopoiesis and leukemia evolution. *Nature Genetics*, 2016.

[36] E Sharon, LV Sibener, A Battle, HB Fraser, KC Garcia, and JK Pritchard. Genetic variation in MHC proteins is associated with T cell receptor expression biases. *Nature Genetics*, 2016.

[37] CC Parker, S Gopalakrishnan, P Carbonetto, NM Gonzales, E Leung, YJ Park, E Aryee, J Davis, DA Blizard, CL Ackert-Bicknell, LA, JK Pritchard, and Palmer AA. Genome-wide association study of behavioral, physiological and gene expression traits in outbred CFW mice. *Nature Genetics*, 2016.

[38] C Kontur, S Kumar, X Lan, JK Pritchard, and AP Turkewitz. Whole genome sequencing identifies a novel factor required for secretory granule maturation in Tetrahymena thermophila. *G3*, 6:2505–2516, 2016.

[39] A Raj, SH Wang, H Shim, A Harpak, YI Li, B Engelmann, M Stephens, Y Gilad, and JK Pritchard. Thousands of novel translated open reading frames in humans inferred by ribosome footprint profiling. *eLife*, 5:e13328, 2016.

[40] X Lan and JK Pritchard. Coregulation of tandem duplicate genes slows evolution of subfunctionalization in mammals. *Science*, 352(6288):1009–1013, 2016.

[41] YI Li, B van de Geijn, A Raj, DA Knowles, AA Petti, D Golan, Y Gilad, and JK Pritchard. RNA splicing is a primary link between genetic variation and disease. *Science*, 352(6285):600–604, 2016.

[42] CK Burrows, NE Banovich, BJ Pavlovic, K Patterson, I Gallego Romero, JK Pritchard, and Y Gilad. Genetic variation, not cell type of origin, underlies the majority of identifiable regulatory differences in iPSCs. *PLoS Genet*, 12(1):e1005793, 2016.

[43] M Gymrek, T Willems, A Guilmatre, H Zeng, B Markus, S Georgiev, MJ Daly, AL Price, JK Pritchard, AJ Sharp, and Y Erlich. Abundant contribution of short tandem repeats to gene expression variation in humans. *Nature Genetics*, 48:22–29, 2016.

[44] A Raj, H Shim, Y Gilad, JK Pritchard, and M Stephens. msCentipede: Modeling heterogeneity across genomic sites and replicates improves accuracy in the inference of transcription factor binding. *PLOS One*, 10(9):e0138030, 2015.

[45] B van de Geijn, G McVicker, Y Gilad, and JK Pritchard. WASP: allele-specific software for robust molecular quantitative trait locus discovery. *Nature Methods*, 12(11):1061–1063, 2015.

[46] Grubert F, Zaugg JB, Kasowski M, Ursu O, Spacek DV, Martin AR, Greenside P, Srivas R, Phanstiel DH, Pekowska A, Heidari N, Euskirchen G, Huber W, Pritchard JK, Bustamante CD, Steinmetz LM, Kundaje A, and Snyder M. Genetic control of chromatin states in humans involves local and distal chromosomal interactions. *Cell*, 162(5):1051–1065, 2015.

[47] SM Thomas, C Kagan, BJ Pavlovic, J Burnett, K Patterson, JK Pritchard, and Y Gilad. Reprogramming LCLs to iPSCs results in recovery of donor-specific gene expression signature. *PLOS Genet.*, 11(5):e1005216, 2015.

[48] A Battle, Z Khan, SH Wang, A Mitrano, MJ Ford, JK Pritchard, and Y Gilad. Impact of regulatory variation from RNA to protein. *Science*, 347:664–667, 2014.

[49] AA Pai, JK Pritchard, and Y Gilad. The genetic and mechanistic basis for variation in gene regulation. *PLOS Genetics*, 11(1):e1004857, 2015.

[50] X Zhou, C Cain, M Myrthil, N Lewellen, K Michelini, E Davenport, M Stephens, JK Pritchard, and Y Gilad. Epigenetic modifications are associated with inter-species gene expression variation in primates. *Genome Biology*, 15(12):547, 2014.

[51] NE Banovich, X Lan, G McVicker, B Van de Geijn, JF Degner, JD Blischak, JK Pritchard, and Y Gilad. Methylation QTLs are associated with coordinated changes in transcription factor binding, histone modifications, and gene expression levels. *PLOS Genet.*, 10(9):e1004663, 2014.

[52] M Çalışkan, JK Pritchard, C Ober, and Y Gilad. The effect of freeze-thaw cycles on gene expression levels in lymphoblastoid cell lines. *PLOS One*, 9(9):e107166, 2014.

[53] A Raj, M Stephens, and JK Pritchard. Variational inference of population structure in large SNP datasets. *Genetics*, 197:573–589, 2014.

[54] DA Cusanovich, B Pavlovic, JK Pritchard, and Y Gilad. The functional consequences of variation in transcription factor binding. *PLoS Genetics*, 10(3):e1004226, 2014.

[55] C Jeong, G Alkorta-Aranburu, B Basnyat, M Neupane, DB Witonsky, JK Pritchard, CM Beall, and A Di Rienzo. Admixture facilitates genetic adaptations to high altitude in Tibet. *Nature Communications*, 5:3281, 2014.

[56] YB Simons, MC Turchin, JK Pritchard, and G Sella. The deleterious mutation load is insensitive to recent population history. *Nature Genetics*, 46:220–224, 2014.

[57] N Nalabothula, G McVicker, J Maiorano, R Martin, JK Pritchard, and YN Fondufe-Mittendorf. The chromatin architectural proteins HMGD1 and H1 bind reciprocally and have opposite effects on chromatin structure and gene regulation. *BMC Genomics*, 15(1):92, 2014.

[58] E Birney and JK Pritchard. Archaic humans: Four makes a party. *Nature*, 505:32–34, 2014.

[59] G McVicker, B van de Geijn, JF Degner, CE Cain, NE Banovich, A Raj, N Lewellen, M Myrthil, Y Gilad, and JK Pritchard. Identification of genetic variants that affect histone modifications in human cells. *Science*, 342(6159):747–749, 2013.

[60] Z Khan, MJ Ford, DA Cusanovich, A Mitrano, JK Pritchard, and Y Gilad. Primate transcript and protein expression levels evolve under compensatory selection pressures. *Science*, 342(6162):1100–1104, 2013.

[61] G Alkorta-Aranburu, CM Beall, DB Witonsky, A Gebremedhin, JK Pritchard, and A Di Rienzo. The genetic architecture of adaptations to high altitude in Ethiopia. *PLoS Genetics*, 8(12):e1003110, 2012.

[62] DJ Gaffney, G McVicker, AA Pai, YN Fondufe-Mittendorf, N Lewellen, K Michelini, J Widom, Y Gilad, and JK Pritchard. Controls of nucleosome positioning in the human genome. *PLOS Genetics*, 8(11):e1003036, 2012.

[63] JK Pickrell and JK Pritchard. Inference of population splits and mixtures from genome-wide allele frequency data. *PLOS Genetics*, 8(11):e1002967, 2012.

[64] AA Pai, CE Cain, O Mizrahi-Man, S De Leon, N Lewellen, J-B Veyrieras, JF Degner, DJ Gaffney, JK Pickrell, M Stephens, JK Pritchard, and Y Gilad. The contribution of RNA decay quantitative trait loci to inter-individual variation in steady-state gene expression levels. *PLOS Genetics*, 8(10):e1003000, 2012.

[65] J.R. Ecker, W.A. Bickmore, I. Barroso, J.K. Pritchard, Y. Gilad, and E. Segal. Genomics: ENCODE explained. *Nature*, 489(7414):52–55, 2012.

[66] JF Degner, AA Pai, R Pique-Regi, JB Veyrieras, DJ Gaffney, JK Pickrell, S De Leon, K Michelini, N Lewellen, GE Crawford, M Stephens, Y Gilad, and JK Pritchard. DNaseI sensitivity QTLs are a major determinant of human expression variation. *Nature*, 482(7385):390–394, 2012.

[67] JK Pickrell, Y Gilad, and JK Pritchard. Comment on "Widespread RNA and DNA sequence differences in the human transcriptome". *Science*, 335(6074):1302–1302, 2012.

[68] JM Zullo, IA Demarco, R Piqué-Regi, DJ Gaffney, CB Epstein, CJ Spooner, TR Luperchio, BE Bernstein, JK Pritchard, KL Reddy, and H Singh. DNA sequence-dependent compartmentalization and silencing of chromatin at the nuclear lamina. *Cell*, 149(7):1474–1487, 2012.

[69] DJ Gaffney, J-B Veyrieras, JF Degner, PR Roger, AA Pai, GE Crawford, M Stephens, Y Gilad, and JK Pritchard. Dissecting the regulatory architecture of gene expression QTLs. *Genome Biology*, 13(1):R7, 2012.

[70] J.B. Veyrieras, D.J. Gaffney, J.K. Pickrell, Y Gilad, M Stephens, and JK Pritchard. Exon-specific QTLs skew the inferred distribution of expression QTLs detected using gene expression array data. *PLoS ONE*, 7(2):e30629, 2012.

[71] DG MacArthur, S Balasubramanian, A Frankish, N Huang, J Morris, K Walter, L Jostins, L Habegger, JK Pickrell, ...[37 authors]..., Pritchard JK, Barrett JC, Harrow J, Hurles ME, Gerstein MB, and Tyler-Smith C. A systematic survey of loss-of-function variants in human protein-coding genes. *Science*, 335(6070):823–828, 2012.

[72] GH Perry, P Melsted, JC Marioni, Y Wang, R Bainer, JK Pickrell, K Michelini, S Zehr, AD Yoder, M Stephens, JK Pritchard, and Y Gilad. Comparative RNA sequencing reveals substantial genetic variation in endangered primates. *Genome Research*, 22(4):602–610, 2012.

[73] GH Perry, D Reeves, P Melsted, A Ratan, W Miller, K Michelini, EE Louis Jr, JK Pritchard, CE Mason, and Y Gilad. A genome sequence resource for the aye-aye (Daubentonia madagascariensis), a nocturnal lemur from Madagascar. *Genome Biology and Evolution*, 4(2):126–135, 2012.

[74] L. Huang, M. Jakobsson, T.J. Pemberton, M. Ibrahim, T. Nyambo, S. Omar, J.K. Pritchard, S.A. Tishkoff, and N.A. Rosenberg. Haplotype variation and genotype imputation in African populations. *Genetic Epidemiology*, 35(8):766–780, 2011.

[75] JK Pritchard. Whole-genome sequencing data offer insights into human demography. *Nature Genetics*, 43(10):923–925, 2011.

[76] P Melsted and JK Pritchard. Efficient counting of k-mers in DNA sequences using a Bloom filter. *BMC Bioinformatics*, 12(1):333, 2011.

[77] JK Pickrell, DJ Gaffney, Y Gilad, and JK Pritchard. False positive peaks in ChIP-seq and other sequencing-based functional assays caused by unannotated high copy number regions. *Bioinformatics*, 27(15):2144–2146, 2011.

[78] AM Hancock, DB Witonsky, G Alkorta-Aranburu, CM Beall, A Gebremedhin, R Sukernik, G Utermann, JK Pritchard, G Coop, and A Di Rienzo. Adaptations to climate-mediated selective pressures in humans. *PLoS Genetics*, 7(4):e1001375, 2011.

[79] R Pique-Regi, JF Degner, AA Pai, DJ Gaffney, Y Gilad, and JK Pritchard. Accurate inference of transcription factor binding from DNA sequence and chromatin accessibility data. *Genome Research*, 21(3):447–455, 2011.

[80] JT Bell, AA Pai, JK Pickrell, DJ Gaffney, R Pique-Regi, JF Degner, Y Gilad, and JK Pritchard. DNA methylation patterns associate with genetic and gene expression variation in HapMap cell lines. *Genome Biology*, 12(1):R10, 2011.

[81] AA Pai, JT Bell, JC Marioni, JK Pritchard, and Y Gilad. A Genome-Wide Study of DNA Methylation Patterns and Gene Expression Levels in Multiple Human and Chimpanzee Tissues. *PLoS Genetics*, 7(2), 2011.

[82] JK Pickrell, JC Marioni, AA Pai, JF Degner, BE Engelhardt, E Nkadori, JB Veyrieras, M Stephens, Y Gilad, and JK Pritchard. Understanding mechanisms underlying human gene expression variation with RNA sequencing. *Nature*, 464(7289):768–772, 2010.

[83] JK Pickrell, AA Pai, Y Gilad, and JK Pritchard. Noisy splicing drives mRNA isoform diversity in human cells. *PLoS Genet*, 6(12):e1001236, 2010.

[84] JK Pritchard and A Di Rienzo. Adaptation–not by sweeps alone. *Nature Reviews Genetics*, 11(10):665–667, 2010.

[85] JK Pritchard. How we are evolving. *Scientific American*, 301(10):41–47, 2010.

[86] JK Pritchard, JK Pickrell, and G Coop. The genetics of human adaptation: hard sweeps, soft sweeps, and polygenic adaptation. *Current Biology*, 20(4):R208–R215, 2010.

[87] G Coop, D Witonsky, A Di Rienzo, and JK Pritchard. Using environmental correlations to identify loci underlying local adaptation. *Genetics*, 185(4):1411–1423, 2010.

[88] AM Hancock, DB Witonsky, E Ehler, G Alkorta-Aranburu, C Beall, A Gebremedhin, R Sukernik, G Utermann, JK Pritchard, G Coop, and A Di Rienzo. Human adaptations to diet, subsistence, and ecoregion are due to subtle shifts in allele frequency. *Proceedings of the National Academy of Sciences*, 107(Supplement 2):8924–8930, 2010.

[89] Y Gilad, JK Pritchard, and K Thornton. Characterizing natural variation using next-generation sequencing technologies. *Trends in Genetics*, 25(10):463–471, 2009.

[90] JF Degner, JC Marioni, AA Pai, JK Pickrell, E Nkadori, Y Gilad, and JK Pritchard. Effect of read-mapping biases on detecting allele-specific expression from RNA-sequencing data. *Bioinformatics*, 25(24):3207–3212, 2009.

[91] MJ Hubisz, D Falush, M Stephens, and JK Pritchard. Inferring weak population structure with the assistance of sample group information. *Molecular Ecology Resources*, 9(5):1322–1332, 2009.

[92] G Coop, JK Pickrell, J Novembre, S Kudaravalli, J Li, D Absher, RM Myers, LL Cavalli-Sforza, MW Feldman, and JK Pritchard. The role of geography in human adaptation. *PLoS Genetics*, 5(6):e1000500, 2009.

[93] D Davison, JK Pritchard, and G Coop. An approximate likelihood for genetic data under a model with recombination and population splitting. *Theoretical Population Biology*, 75(4):331–345, 2009.

[94] JK Pickrell, G Coop, J Novembre, S Kudaravalli, JZ Li, D Absher, BS Srinivasan, GS Barsh, RM Myers, MW Feldman, and JK Pritchard. Signals of recent positive selection in a worldwide sample of human populations. *Genome Research*, 19(5):826–837, 2009.

[95] KB Schroeder, M Jakobsson, MH Crawford, TG Schurr, SM Boca, DF Conrad, RY Tito, LP Osipova, LA Tarskaia, SI Zhadanov, JD Wall, JK Pritchard, RS Malhi, DG Smith, and NA Rosenberg. Haplotypic background of a private allele at high frequency in the Americas. *Molecular Biology and Evolution*, 26(5):995–1016, 2009.

[96] S Kudaravalli, JB Veyrieras, BE Stranger, ET Dermitzakis, and JK Pritchard. Gene expression levels are a target of recent natural selection in the human genome. *Molecular Biology and Evolution*, 26(3):649–658, 2009.

[97] JB Veyrieras, S Kudaravalli, SY Kim, ET Dermitzakis, Y Gilad, M Stephens, and JK Pritchard. High-resolution mapping of expression-QTLs yields insight into human gene regulation. *PLoS Genet*, 4(10):e1000214, 2008.

[98] Y Gilad, SA Rifkin, and JK Pritchard. Revealing the architecture of gene regulation: the promise of eQTL studies. *Trends in Genetics*, 24(8):408–415, 2008.

[99] G Coop, X Wen, C Ober, JK Pritchard, and M Przeworski. High-resolution mapping of crossovers reveals extensive variation in fine-scale recombination patterns among humans. *Science*, 319(5868):1395–1398, 2008.

[100] AM Hancock, DB Witonsky, AS Gordon, G Eshel, JK Pritchard, G Coop, and A Di Rienzo. Adaptations to climate in candidate genes for common metabolic disorders. *PLoS Genetics*, 4(2):e32, 2008.

[101] TJ Pemberton, M Jakobsson, DF Conrad, G Coop, JD Wall, JK Pritchard, PI Patel, and NA Rosenberg. Using population mixtures to optimize the utility of genomic databases: linkage disequilibrium and association study design in India. *Annals of Human Genetics*, 72(4):535–546, 2008.

[102] SY Kim and JK Pritchard. Adaptive evolution of conserved noncoding elements in mammals. *PLoS Genet*, 3(9):e147, 2007.

[103] J Novembre, JK Pritchard, and G Coop. Adaptive drool in the gene pool. *Nature Genetics*, 39(10):1188–1190, 2007.

[104] EE Eichler, [11 authors], JK Pritchard, and [5 authors]. Completing the map of human genetic variation. *Nature*, 447:161–165, 2007.

[105] S Zöllner and JK Pritchard. Overcoming the winner's curse: estimating penetrance parameters from case-control data. *The American Journal of Human Genetics*, 80(4):605–615, 2007.

[106] D Falush, M Stephens, and JK Pritchard. Inference of population structure using multilocus genotype data: dominant markers and null alleles. *Molecular Ecology Notes*, 7(4):574–578, 2007.

[107] SA Tishkoff, FA Reed, A Ranciaro, BF Voight, CC Babbitt, JS Silverman, K Powell, HM Mortensen, JB Hirbo, M Osman, M Ibrahim, SA Omar, G Lema, TB Nyambo, J Ghori, S Bumpstead, JK Pritchard, GA Wray, and P Deloukas. Convergent adaptation of human lactase persistence in Africa and Europe. *Nat Genet*, 39:31–40, 2007.

[108] JP Noonan, G Coop, S Kudaravalli, D Smith, J Krause, J Alessi, F Chen, D Platt, S Pääbo, JK Pritchard, and EM Rubin. Sequencing and analysis of Neanderthal genomic DNA. *Science*, 314:1113–1118, 2006.

[109] DF Conrad, M Jakobsson, G Coop, X Wen, JD Wall, NA Rosenberg, and JK Pritchard. A worldwide survey of haplotype variation and linkage disequilibrium in the human genome. *Nature Genetics*, 38:1251–1260, 2006.

[110] C Murgia, JK Pritchard, SY Kim, A Fassati, and RA Weiss. Clonal origin and evolution of a transmissible cancer. *Cell*, 126:477–487, 2006.

[111] BF Voight, S Kudaravalli, X Wen, and JK Pritchard. A map of recent positive selection in the human genome. *Plos Biology*, 4(3):e72, 2006.

[112] DF Conrad, TD Andrews, NP Carter, ME Hurles, and JK Pritchard. A high-resolution survey of deletion polymorphism in the human genome. *Nature Genetics*, 38:75–81, 2006.

[113] G Hellenthal, JK Pritchard, and M Stephens. The effects of genotype-dependent recombination, and transmission asymmetry, on linkage disequilibrium. *Genetics*, 172:2001–2005, 2006.

[114] NA Rosenberg, S Mahajan, S Ramachandran, C Zhao, JK Pritchard, and MW Feldman. Clines, clusters, and the effect of study design on the inference of human population structure. *PLoS Genetics*, 1(6):e70, 2005.

[115] BF Voight and JK Pritchard. Confounding from cryptic relatedness in case-control association studies. *PLoS Genetics*, 1(e32):1–10, 2005.

[116] S Zöllner and JK Pritchard. Coalescent-based association mapping and fine mapping of complex trait loci. *Genetics*, 169:1071–1092, 2005.

[117] S Zollner, X Wen, and JK Pritchard. Association mapping and fine mapping with TreeLD. *Bioinformatics*, 21:3168–70, 2005.

[118] G Montana and JK Pritchard. Statistical tests for admixture mapping with case-control and case-only data. *Am. J. Hum. Genet.*, 75:771–789, 2004.

[119] S Zöllner and JK Pritchard. A coalescent-based approach for complex disease mapping. *Lecture notes in bioinformatics: Computational methods for SNPs and haplotype inference*, 2983:124–130, 2004.

[120] S Zöllner, X Wen, NA Hanchard, MA Herbert, C Ober, and JK Pritchard. Genome-wide evidence for extensive transmission distortion in humans. *Am. J. Hum. Genet.*, 74:62–72, 2004.

[121] NA Rosenberg, L Lei, R Ward, and JK Pritchard. Informativeness of genetic markers for inference of ancestry. *Am. J. Hum. Genet.*, 73:1402–1422, 2003.

[122] JD Wall and JK Pritchard. Assessing the performance of the haplotype block model of linkage disequilibrium. *Am. J. Hum. Genet.*, 73:502–515, 2003.

[123] JD Wall and JK Pritchard. Haplotype blocks and the structure of linkage disequilibrium in the human genome. *Nature Reviews Genetics*, 4:587–597, 2003.

[124] NA Rosenberg, JK Pritchard, JL Weber, HM Cann, KK Kidd, LA Zhivotovsky, and MW Feldman. Response to comment on "Genetic structure of human populations". *Science*, 300:1877, 2003.

[125] D Falush, M Stephens, and JK Pritchard. Inference of population structure: Extensions to linked loci and correlated allele frequencies. *Genetics*, 164:1567–1587, 2003.

[126] D Falush, T Wirth, B Linz, JK Pritchard, M Stephens, and [13 others]. Traces of human migrations in *Helicobacter pylori* populations. *Science*, 299:1582–1585, 2003.

[127] NA Rosenberg, JK Pritchard, JL Weber, HM Cann, KK Kidd, LA Zhivotovsky, and MW Feldman. Genetic structure of human populations. *Science*, 298:2381–5, 2002.

[128] JK Pritchard and NJ Cox. The allelic architecture of human disease genes: common disease–common variant...or not? *Human Molecular Genetics*, 11:2417–2423, 2002.

[129] JK Pritchard and P Donnelly. Case-control studies of association in structured or admixed populations. *Theor Popul Biol*, 60:227–37, 2001.

[130] JK Pritchard. Are rare variants responsible for susceptibility to common diseases? *Am. J. Hum. Genet.*, 69:124–137, 2001.

[131] JK Pritchard. Deconstructing maize population structure. *Nature Genetics*, 28:203–204, 2001.

[132] JK Pritchard and M Przeworski. Linkage disequilibrium in humans: models and data. *Am. J. Hum. Genet.*, 69:1–14, 2001.

[133] NA Rosenberg, E Woolf, JK Pritchard, T Schaap, D Gefel, I Shpirer, U Lavi, B Bonne-Tamir, J Hillel, and MW Feldman. Distinctive genetic signatures in the Libyan Jews. *Proc Natl Acad Sci U S A*, 98:858–63, 2001.

[134] R Thompson, JK Pritchard, P Shen, PJ Oefner, and MW Feldman. Recent common ancestry of human Y chromsomes: Evidence from DNA sequence data. *Proc. Natl. Acad. Sci. USA*, 97:7360–7365, 2000.

[135] M Beaumont, D Gottelli, EM Barratt, AC Kitchener, MJ Daniels, JK Pritchard, and MW Bruford. Genetic diversity and introgression in the Scottish wildcat. *Molecular Ecology*, 10:319–336, 2001.

[136] JK Pritchard, M Stephens, NA Rosenberg, and P Donnelly. Association mapping in structured populations. *Am. J. Hum. Genet.*, 67:170–181, 2000.

[137] JK Pritchard, M Stephens, and P Donnelly. Inference of population structure using multilocus genotype data. *Genetics*, 155:945–959, 2000.

[138] JK Pritchard and NA Rosenberg. Use of unlinked genetic markers to detect population stratification in association studies. *Am. J. Hum. Gen.*, 65:220–228, 1999.

[139] JK Pritchard, MT Seielstad, A Perez-Lezaun, and MW Feldman. Population growth of human Y chromosomes: a study of Y chromosome microsatellites. *Mol. Biol. and Evol.*, 16:1791–1798, 1999.

[140] MW Feldman, J Kumm, and JK Pritchard. Mutation and migration in models of microsatellite evolution. In D.G. Goldstein and C. Schlotterer, editors, *Microsatellites: Evolution and Applications*, pages 98–115. Oxford University Press, Oxford, UK., 1999.

[141] AJ Robson, CT Bergstrom, and JK Pritchard. Risky business: sexual and asexual reproduction in variable environments. *J Theor Biol*, 197:541–56, 1999.

[142] JK Pritchard and MW Feldman. A test for heterogeneity of microsatellite variation. In M. K. Uyenoyama and A. von Haeseler, editors, *Proceedings of the Trinational Workshop on Molecular Evolution*, pages 47–56. Duke University Publications Group, Durham, NC, 1998.

[143] CT Bergstrom and JK Pritchard. Germline bottlenecks and the evolutionary maintenance of mitochondrial genomes. *Genetics*, 149:2135–2146, 1998.

[144] JK Pritchard and SW Schaeffer. Polymorphism and divergence at a $Drosophila\ melanogaster$ pseudogene locus. *Genetics*, 147:199–208, 1997.

[145] JK Pritchard and MW Feldman. Statistics for microsatellite variation based on coalescence. *Theor. Pop. Biol.*, 50:325–344, 1996.

[146] JK Pritchard and MW Feldman. Genetic data and the African origin of modern humans [technical comment]. *Science*, 274:1548, 1996.

[147] WG Pritchard and JK Pritchard. Mathematical models of running. *American Scientist*, 82:546–553, 1994.