JONATHAN K. PRITCHARD
CURRICULUM VITAE

Professor, Departments of Biology and Genetics
Stanford University
Investigator, Howard Hughes Medical Institute

Mail: Department of Genetics, Mail Stop-5120
Stanford, California 94305-5120
Web: http://pritchardlab.stanford.edu
Email: pritch@stanford.edu
CV updated: November 2015

Employment History

2013–Present	Professor, Departments of Genetics and Biology, Stanford University
2008–Present	Investigator, Howard Hughes Medical Institute
2006–2013	Professor, The University of Chicago
2001–2005	Assistant Professor, The University of Chicago Department of Human Genetics Committee on Genetics, Genomics and Systems Biology Committee on Evolutionary Biology
1998–2001	Postdoctoral Fellow, Statistics University of Oxford Advised by Peter Donnelly

Education

1994–1998	Ph.D., Biology Stanford University Advised by Marcus Feldman
1989–1994	B.Sc., Biology and Mathematics Pennsylvania State University

Research Support

2008–present	Investigator, Howard Hughes Medical Institute.
2012-2015	U01 HG007036 (PI): "Genetic and epigenetic controls of gene regulation"
2014-2016	RO1 ES025009 (PI): "Computational methods for modeling lineage-specific gene regulation"
2012-2017	RO1 MH084703 (Co-PI): "Analysis and interpretation of noncoding regulatory variation"

Previous grants/fellowships:

2010-2013	RO1 MH090951: "Statistical analysis of gene expression quantitative trait loci (eQTL)" includes administrative supplement for 2012-2013
2008-2011	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"
1998	NSF funding to spend three months at Isaac Newton Institute, Cambridge, UK
1997	NSF travel award to TriNational Workshop on Molecular Evolution, Munich
1995	Santa Fe Institute Complex Systems Summer School, fellowship
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

Awards and honors

2015	Stanford 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 G. meeting
2008	Selected as an investigator of the Howard Hughes Medical Institute
2007	*Best postdoc talk (basic research)* to my postdoc Graham Coop at Am. Soc. Hum. Genet. mtg.
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 in humans
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." Awarded for the *Genetics* paper of Pritchard et al (2000) on population structure.
1999-2003	Burroughs-Wellcome Fund Hitchings-Elion Postdoc-Faculty Award
1994-1998	Howard Hughes Medical Institute Predoctoral Fellowship
1994	NSF Predoctoral Fellowship (declined)
1994	McCoy Award, given to the Penn State Outstanding Scholar-Athlete
1989-1993	Penn State Braddock Scholarship (four-year full-tuition award)
1989-1993	Penn State Academic Excellence Scholarship
1992	Stecker Award, given to a Penn State undergraduate in Mathematics

Selected Professional Activities

2013–present	Classroom teaching at Stanford: *Statistical and Machine Learning Methods for Genomics* with Hua Tang, Anshul Kundaje miscellaneous lectures in other classes
2013–present	Member of various Stanford internal committees in Biology and Genetics:

	many thesis committees; promotions; ...
2001–present	Member of various U of C internal committees (e.g., graduate admissions, graduate curriculum committee, many student thesis committees, BSD Cmte on Academic Promotions, various search committees, University Board of Academic Computing)
2007–present	Instructor at Conservation Genetics Workshop, Flathead Lake MT (biennial)
2001–present	Reviewer for NIH, NSF, HHMI, Wellcome Trust, etc (on an *ad hoc* basis)
2006–present	External referee for many promotion/tenure cases
1998–present	Reviewer for many journals (Nature, Science, Cell, PNAS, PLoS, etc)
2012–present	Computational Advisory Board, DNAnexus
2007–present	Scientific Advisory Board, 23AndMe
2013–present	Scientific Advisory Board, Cellular Genomics Program, Wellcome Trust Sanger Institute
2001–2013	Classroom teaching at University of Chicago: ∼15-30 lecture hours per year: *Human Variation and Disease* (grad; 2002-present); *Simulation and Modeling of Evolutionary Processes* (grad/undergrad; 2006-2009); *AP5 Biology* (Freshman advanced biology; 2012, 2013); smaller contributions to various other classes.
2016–present	NHGRI (NIH) Council Member (appointment pending)
2016–present	Lead organizing committee for Biology of Genomes, Cold Spring Harbor
2015–present	Science Magazine Board of Reviewing Editors
2014–present	Frequent guest editor for PLOS Genetics
2012–present	Editorial Board: Current Biology
2006–2014	Associate Editor: PLOS Genetics
2008–2012	Associate Editor: Molecular Biology and Evolution

Graduate Student/Postdoc Mentoring:

2015–Present	Kelley Harris (postdoc)
2015–Present	Ziyue Gao (postdoc)
2015–Present	David Knowles (postdoc); joint with Sylvia Plevritis
2014–Present	Yang Li (postdoc)
2014–Present	Anand Bhaskar (postdoc)
2014–Present	David Golan (postdoc)
2014–Present	Eilon Sharon (postdoc); joint with Hunter Fraser
2012–Present	Yair Field (postdoc)
2012–Present	Xun Lan (postdoc)
2011–Present	Anil Raj (postdoc)
2015–present	Jessica Ribado (PhD student, Genetics; joint with Ami Bhatt)
2015–present	Evan Boyle (PhD student, Genetics; joint with Will Greenleaf)
2014–present	Diego Calderon (PhD student, BMI)
2014–present	Emily Glassberg (PhD student, Biology)
2014–present	Arbel Harpak (PhD student, Biology)
2014–present	Natalie Telis (PhD student, BMI)
2011–Present	Bryce van der Geijn (PhD student, Chicago GGSB)
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 assistant professor, Johns Hopkins
2011–2014	Stoyan Georgiev (postdoc); now data scientist, Google
2011–2013	Zia Khan (postdoc; joint with Y Gilad); now assistant professor, University of Maryland
2011–2013	Shyam Gopalakrishnan (postdoc; joint with Abe Palmer); now researcher 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 assistant professor, Wayne State, MI
2008–2011	Daniel Gaffney (postdoc); now junior group leader, Sanger Institute, UK
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 associate professor, U. of California, Davis
2001–2005	Sebastian Zöllner (postdoc); now associate 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
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; now asst prof NY Genome Center/Columbia Univ
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
2003–2007	Donald Conrad (PhD, Human Genetics, 2008) postdoc with Matt Hurles, Sanger Institute; now asst. prof. Wash U.
2002–2006	Benjamin Voight (PhD, Human Genetics, 2006; joint with N. Cox) postdoc with Mark Daly, Broad Institute; now asst. prof., Univ of Pennsylvania
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
2011-2012	Andy Dahl (BSc & MSc, Statistics); now DPhil student, Oxford
2006–2008	Melissa Hubisz (M.S., Human Genetics); now PhD student, Cornell

Research Overview

A central problem facing modern human genetics is to make sense of the vast quantity of human genetic variation. Which of the ∼10 million common SNPs and thousands of deletions and duplications contribute to complex diseases or other traits? How important are rare variants in driving phenotypic variation? How can we identify the functional variants that underlie observed disease associations? What impact do purifying selection and adaptation have on genetic differences within and between populations, or between different species?

My group is interested in these and similar questions relating to the causes and consequences of genetic variation. Our work is primarily computational, with emphasis on data analysis, development of statistical methods and modeling–often working closely with experimental groups. Recently we have become interested in understanding how genetic variation within and between species impacts gene regulation, and this is now a major part of our research, in close collaboration with Yoav Gilad's group at the University of Chicago.

Our past research contributions and current interests include the following:

- **Inference of population structure.** My colleagues and I have developed a model-based clustering approach (named *structure*) for using multilocus genotype data to infer population structure, and assign individuals to populations [94, 82, 63, 48]. This method is now widely used in a range of fields, including in human genetics, forensics, and perhaps most widely in molecular ecology and conservation genetics. In 2002, our original paper [94] won the Mitchell Prize from the International Society for Bayesian Applications and it has been highly cited (> 9000 times according to Google Scholar). Our application to human data [84] won Lancet's Paper of the Year award in 2003.

- **Correcting for population structure in association mapping.** Case-control association mapping is widely perceived as a powerful tool for gene identification, but this approach fell out of favor in the human genetics community in the 1990s due to concern that unrecognized population structure could lead to false positives. Our work in this area helped to show that these problems can be greatly reduced by using random unlinked marker loci to detect, and correct for cryptic population structure [95, 93, 86]. (Similar ideas were developed independently by Bernie Devlin and Kathryn Roeder and slightly later by David Reich and colleagues.) The case-control design is now the most widely used approach in the field, and virtually all genome-wide association studies now use some sort of test for population structure.

  In a separate line of work, in 2001 I used population genetic arguments to investigate the likely role of rare variants in complex diseases; this has important implications for the likely success of different types of study design [87, 85]. We have followed this up recently [13] with further analysis arguing that rare variants are likely to be important for traits that are closely tied to fitness, but otherwise not.

- **Human variation and history.** I have a long-standing interest in using patterns of genetic variation to learn about evolutionary or historical processes, in addition to our work on human population structure. My 1999 paper on inferring human demographic history from Y chromosome data [96] was among the first papers to apply Approximate Bayesian Computation in genetics. My lab provided the statistical analysis for the first successful autosomal sequencing of Neanderthal DNA [65]. We performed one of the first high-resolution surveys of deletions in the human genome, showing that copy number variation is extremely (and at the time, surprisingly) widespread [69]. More recently we have developed a new model called TreeMix for jointly inferring population splits and mixture events from genome-wide SNP data [20].

  Related work has considered how demography and the biology of recombination shape linkage disequilibrium in humans [89, 79, 66]. Working with Molly Przeworski, we provided the first pedigree-based approach to studying the genome-wide distribution of recombination events at very high resolution [56]. Our work showed that there was variation across individuals in the extent to which each individual used known recombination hotspots; subsequent work on PRDM9 (by other groups) has helped to define the underlying mechanism of this variation.

- **Adaptation in humans.** We have also put considerable effort into studying the extent of recent natural selection in humans [68, 51, 49, 42, 41, 43, 18]. In our most recent work, we have argued that, while examples of strong, selective sweeps exist, the data suggest that strong, rapid selective sweeps are actually rare in humans [49, 42]. We propose that many adaptive events occur by a polygenic mechanism that allows rapid adaptation of populations, yet may leave relatively little trace in population genetic data [42, 41].

- **Population genetics of gene expression.** Since 2008 a major focus of my lab has been on understanding the links between genetic variation and functional variation in gene regulation [54, 39, 36, 26, 23, 21]. There is now strong evidence that genetic variants that impact gene regulation play important roles in complex traits and in adaptation. Yet we know relatively little about the detailed mechanisms by

which variation impacts expression, or how to predict which SNPs are likely to affect expression in any given cell type.

We are now working on an intensive experimental and computational project in this area, in close collaboration with our colleague Yoav Gilad and his lab. At present much of our effort is going into an in-depth study of the variants (and mechanisms) that generate expression QTLs in the HapMap lymphoblastoid cell lines. These cells represent a unique resource for our purpose, as they have been sequenced by the 1000 Genomes Project, and characterized using a variety of genomic technologies. In this project we are developing statistical and computational methods to integrate diverse types of genome-wide information to better understand the links between genetic variation and gene expression. In recent work we have been able to link about half of all eQTLs to mechanisms involving changes in chromatin accessibility [23], and a smaller fraction to changes in mRNA decay rates [21].

We are continuing to work on LCLs, as well as expanding to other cell types through iPSC approaches and through our involvement in the GTEx Consortium. Ultimately we hope to gain much better understanding of the mechanisms by which genetic variation impacts gene regulation in a much broader range of cell types; we hope that this type of work will be of great value for interpreting population variation data.

Publications

[1] 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.

[2] 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.

[3] 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.

[4] 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.

[5] 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.

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

[7] 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.

[8] 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.

[9] 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.

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

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

[12] 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, 2014.

[13] 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.

[14] 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.

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

[16] 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.

[17] 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.

[18] Gorka Alkorta-Aranburu, Cynthia M Beall, David B Witonsky, Amha Gebremedhin, Jonathan K Pritchard, and Anna Di Rienzo. The genetic architecture of adaptations to high altitude in Ethiopia. *PLoS Genetics*, 8(12):e1003110, 2012.

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

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

[21] A.A. Pai, C.E. Cain, O. Mizrahi-Man, S. De Leon, N. Lewellen, J.-B. Veyrieras, J.F. Degner, D.J. Gaffney, J.K. Pickrell, M. Stephens, J.K. 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.

[22] 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.

[23] J.F. Degner, A.A. Pai, R. Pique-Regi, J.B. Veyrieras, D.J. Gaffney, J.K. Pickrell, S. De Leon, K. Michelini, N. Lewellen, G.E. Crawford, M. Stephens, Y. Gilad, and J.K. Pritchard. DNaseI sensitivity QTLs are a major determinant of human expression variation. *Nature*, 482(7385):390–394, 2012.

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

[25] J.M. Zullo, I.A. Demarco, R. Piqué-Regi, D.J. Gaffney, C.B. Epstein, C.J. Spooner, T.R. Luperchio, B.E. Bernstein, J.K. Pritchard, K.L. Reddy, and H Singh. DNA sequence-dependent compartmentalization and silencing of chromatin at the nuclear lamina. *Cell*, 149(7):1474–1487, 2012.

[26] D.J. Gaffney, J.B. Veyrieras, J.F. Degner, P.R. Roger, A.A. Pai, G.E. Crawford, M. Stephens, Y. Gilad, and J.K. Pritchard. Dissecting the regulatory architecture of gene expression QTLs. *Genome Biology*, 13(1):R7, 2012.

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

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

[29] G.H. Perry, P. Melsted, J.C. Marioni, Y. Wang, R. Bainer, J.K. Pickrell, K. Michelini, S. Zehr, A.D. Yoder, M. Stephens, J.K. Pritchard, and Y. Gilad. Comparative RNA sequencing reveals substantial genetic variation in endangered primates. *Genome Research*, 22(4):602–610, 2012.

[30] G.H. Perry, D. Reeves, P. Melsted, A. Ratan, W. Miller, K. Michelini, E.E. Louis Jr, J.K. Pritchard, C.E. 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.

[31] 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.

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

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

[34] J.K. Pickrell, D.J. Gaffney, Y. Gilad, and J.K. 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.

[35] A.M. Hancock, D.B. Witonsky, G. Alkorta-Aranburu, C.M. Beall, A. Gebremedhin, R. Sukernik, G. Utermann, J.K. Pritchard, G. Coop, and A. Di Rienzo. Adaptations to climate-mediated selective pressures in humans. *PLoS Genetics*, 7(4):e1001375, 2011.

[36] R. Pique-Regi, J.F. Degner, A.A. Pai, D.J. Gaffney, Y. Gilad, and J.K. Pritchard. Accurate inference of transcription factor binding from DNA sequence and chromatin accessibility data. *Genome Research*, 21(3):447–455, 2011.

[37] J.T. Bell, A.A. Pai, J.K. Pickrell, D.J. Gaffney, R. Pique-Regi, J.F. Degner, Y. Gilad, and J.K. Pritchard. DNA methylation patterns associate with genetic and gene expression variation in HapMap cell lines. *Genome Biology*, 12(1):R10, 2011.

[38] A.A. Pai, J.T. Bell, J.C. Marioni, J.K. 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.

[39] J.K. Pickrell, J.C. Marioni, A.A. Pai, J.F. Degner, B.E. Engelhardt, E. Nkadori, J.B. Veyrieras, M. Stephens, Y. Gilad, and J.K. Pritchard. Understanding mechanisms underlying human gene expression variation with RNA sequencing. *Nature*, 464(7289):768–772, 2010.

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

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

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

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

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

[45] A.M. Hancock, D.B. Witonsky, E. Ehler, G. Alkorta-Aranburu, C. Beall, A. Gebremedhin, R. Sukernik, G. Utermann, J. 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.

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

[47] J.F. Degner, J.C. Marioni, A.A. Pai, J.K. Pickrell, E. Nkadori, Y. Gilad, and J.K. Pritchard. Effect of read-mapping biases on detecting allele-specific expression from RNA-sequencing data. *Bioinformatics*, 25(24):3207–3212, 2009.

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

[49] G. Coop, J.K. Pickrell, J. Novembre, S. Kudaravalli, J. Li, D. Absher, R.M. Myers, L.L. Cavalli-Sforza, M.W. Feldman, and J.K. Pritchard. The role of geography in human adaptation. *PLoS Genetics*, 5(6):e1000500, 2009.

[50] 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.

[51] J.K. Pickrell, G. Coop, J. Novembre, S. Kudaravalli, J.Z. Li, D. Absher, B.S. Srinivasan, G.S. Barsh, R.M. Myers, M.W. Feldman, and J.K. Pritchard. Signals of recent positive selection in a worldwide sample of human populations. *Genome Research*, 19(5):826–837, 2009.

[52] K.B. Schroeder, M. Jakobsson, M.H. Crawford, T.G. Schurr, S.M. Boca, D.F. Conrad, R.Y. Tito, L.P. Osipova, L.A. Tarskaia, S.I. Zhadanov, J.D. Wall, J.K. Pritchard, R.S. Malhi, D.G. Smith, and N.A. Rosenberg. Haplotypic background of a private allele at high frequency in the Americas. *Molecular Biology and Evolution*, 26(5):995–1016, 2009.

[53] S. Kudaravalli, J.B. Veyrieras, B.E. Stranger, E.T. Dermitzakis, and J.K. Pritchard. Gene expression levels are a target of recent natural selection in the human genome. *Molecular Biology and Evolution*, 26(3):649–658, 2009.

[54] J.B. Veyrieras, S. Kudaravalli, S.Y. Kim, E.T. Dermitzakis, Y. Gilad, M. Stephens, and J.K. Pritchard. High-resolution mapping of expression-QTLs yields insight into human gene regulation. *PLoS Genet*, 4(10):e1000214, 2008.

[55] Y. Gilad, S.A. Rifkin, and J.K. Pritchard. Revealing the architecture of gene regulation: the promise of eQTL studies. *Trends in Genetics*, 24(8):408–415, 2008.

[56] G. Coop, X. Wen, C. Ober, J.K. 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.

[57] A.M. Hancock, D.B. Witonsky, A.S. Gordon, G. Eshel, J.K. Pritchard, G. Coop, and A. Di Rienzo. Adaptations to climate in candidate genes for common metabolic disorders. *PLoS Genetics*, 4(2):e32, 2008.

[58] T.J. Pemberton, M. Jakobsson, D.F. Conrad, G. Coop, J.D. Wall, J.K. Pritchard, P.I. Patel, and N.A. 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.

[59] S.Y. Kim and J.K. Pritchard. Adaptive evolution of conserved noncoding elements in mammals. *PLoS Genet*, 3(9):e147, 2007.

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

[61] E.E. Eichler, [11 authors], J.K. Pritchard, and [5 authors]. Completing the map of human genetic variation. *Nature*, 447:161–165, 2007.

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

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

[64] S. A. Tishkoff, F. A. Reed, A. Ranciaro, B. F. Voight, C. C. Babbitt, J. S. Silverman, K. Powell, H. M. Mortensen, J. B. Hirbo, M. Osman, M. Ibrahim, S. A. Omar, G. Lema, T. B. Nyambo, J. Ghori, S. Bumpstead, J. K. Pritchard, G. A. Wray, and P. Deloukas. Convergent adaptation of human lactase persistence in Africa and Europe. *Nat Genet*, 39:31–40, 2007.

[65] J.P. Noonan, G. Coop, S. Kudaravalli, D. Smith, J. Krause, J. Alessi, F. Chen, D. Platt, S. Pääbo, J.K. Pritchard, and E.M. Rubin. Sequencing and analysis of Neanderthal genomic DNA. *Science*, 314:1113–1118, 2006.

[66] D.F. Conrad, M. Jakobsson, G. Coop, X. Wen, J.D. Wall, N.A. Rosenberg, and J.K. Pritchard. A worldwide survey of haplotype variation and linkage disequilibrium in the human genome. *Nature Genetics*, 38:1251–1260, 2006.

[67] C. Murgia, J. K. Pritchard, S.Y. Kim, A. Fassati, and R.A. Weiss. Clonal origin and evolution of a transmissible cancer. *Cell*, 126:477–487, 2006.

[68] B. F. Voight, S. Kudaravalli, X. Wen, and J. K. Pritchard. A map of recent positive selection in the human genome. *Plos Biology*, 4(3):e72, 2006.

[69] D. F. Conrad, T. D. Andrews, N. P. Carter, M. E. Hurles, and J. K. Pritchard. A high-resolution survey of deletion polymorphism in the human genome. *Nature Genetics*, 38:75–81, 2006.

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

[71] N. A. Rosenberg, S. Mahajan, S. Ramachandran, C. Zhao, J. K. Pritchard, and M. W. Feldman. Clines, clusters, and the effect of study design on the inference of human population structure. *PLoS Genetics*, 1(6):e70, 2005.

[72] B. F. Voight and J. K. Pritchard. Confounding from cryptic relatedness in case-control association studies. *PLoS Genetics*, 1(e32):1–10, 2005.

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

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

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

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

[77] S. Zöllner, X. Wen, N. A. Hanchard, M. A. Herbert, C. Ober, and J. K. Pritchard. Genome-wide evidence for extensive transmission distortion in humans. *Am. J. Hum. Genet.*, 74:62–72, 2004.

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

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

[80] J. D. Wall and J. K. Pritchard. Haplotype blocks and the structure of linkage disequilibrium in the human genome. *Nature Reviews Genetics*, 4:587–597, 2003.

[81] N. A. Rosenberg, J. K. Pritchard, J. L. Weber, H. M. Cann, K. K. Kidd, L. A. Zhivotovsky, and M. W. Feldman. Response to comment on "Genetic structure of human populations". *Science*, 300:1877, 2003.

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

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

[84] N. A. Rosenberg, J. K. Pritchard, J. L. Weber, H. M. Cann, K. K. Kidd, L. A. Zhivotovsky, and M. W. Feldman. Genetic structure of human populations. *Science*, 298:2381–5, 2002.

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

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

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

[88] J. K. Pritchard. Deconstructing maize population structure. *Nature Genetics*, 28:203–204, 2001.

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

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

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

[92] M. Beaumont, D. Gottelli, E. M. Barratt, A. C. Kitchener, M. J. Daniels, J. K. Pritchard, and M. W. Bruford. Genetic diversity and introgression in the Scottish wildcat. *Molecular Ecology*, 10:319–336, 2001.

[93] J. K. Pritchard, M. Stephens, N. A. Rosenberg, and P. Donnelly. Association mapping in structured populations. *Am. J. Hum. Genet.*, 67:170–181, 2000.

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

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

[96] J. K. Pritchard, M. T. Seielstad, A. Perez-Lezaun, and M. W. Feldman. Population growth of human Y chromosomes: a study of Y chromosome microsatellites. *Mol. Biol. and Evol.*, 16:1791–1798, 1999.

[97] M. W. Feldman, J. Kumm, and J. K. 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.

[98] A. J. Robson, C. T. Bergstrom, and J. K. Pritchard. Risky business: sexual and asexual reproduction in variable environments. *J Theor Biol*, 197:541–56, 1999.

[99] J. K. Pritchard and M. W. 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.

[100] C. T. Bergstrom and J. K. Pritchard. Germline bottlenecks and the evolutionary maintenance of mitochondrial genomes. *Genetics*, 149:2135–2146, 1998.

[101] J. K. Pritchard and S. W. Schaeffer. Polymorphism and divergence at a $Drosophila\ melanogaster$ pseudogene locus. *Genetics*, 147:199–208, 1997.

[102] J. K. Pritchard and M. W. Feldman. Statistics for microsatellite variation based on coalescence. *Theor. Pop. Biol.*, 50:325–344, 1996.

[103] J. K. Pritchard and M. W. Feldman. Genetic data and the African origin of modern humans [technical comment]. *Science*, 274:1548, 1996.

[104] W. G. Pritchard and J. K. Pritchard. Mathematical models of running. *American Scientist*, 82:546–553, 1994.