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CURRICULUM VITAE

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CV updated: January 2007

Employment History

2006–Present Professor,
2001–2005 Assistant Professor,
Department of Human Genetics
Committees on Genetics and Evolutionary Biology
The University of Chicago

1998–2001 Postdoctoral Fellow, advised by Peter Donnelly
Department of Statistics, University of Oxford

Education

1998 Ph.D., advised by Marcus Feldman
Biological Sciences, Stanford University

1994 B.Sc., Biology (genetics concentration) and Mathematics
Honors project advised by Stephen Schaeffer
Pennsylvania State University

Research Grants

2003-2008 RO1 HG002772: “Linkage Disequilibrium Methods for Complex Trait Mapping”
2004–2009 Packard Foundation Fellowship: “Population genetics of genomic rearrangements”
2005-2009 RO1 DK056670: PI is A. Di Rienzo. “Evolutionary genetics of the metabolic syndrome”

Previous grants/fellowships:

2004-2006 Sloan Foundation Fellowship
1999-2003 Burroughs-Wellcome Fund Hitchings-Elion Postdoc-Faculty Award (\$330K)
“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, etc

- 2006 My student Ben Voight won best Ph.D thesis award in U of C Biological Sciences Division
- 2006 New York Times front page news article on Voight et al paper on selection
- 2004 Selected Packard Fellow (see also Grants)
- 2004 Selected Alfred P. Sloan Fellow (see also Grants)
- 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.
- 1994 McCoy Award, given to the Penn State Outstanding Scholar-Athlete
- 1992 Stecker Award, given to a Penn State undergraduate in Mathematics

Student/Postdoc Mentoring:

- 2004–Present Graham Coop (postdoc)
- 2006–Present Jean-Baptiste Veyrieras (postdoc)
- 2003–Present Donald Conrad (PhD student, Human Genetics)
- 2004–Present Su Yeon Kim (PhD student, Statistics; joint with P. McCullagh)
- 2004–Present Sridhar Kudravalli (PhD student, Human Genetics)
- 2006–Present Melissa Hubisz (PhD student, Human Genetics)

- 2002–2006 Benjamin Voight (PhD student, Human Genetics; joint with N. Cox)
now postdoc with M. Daly, Broad Institute
- 2004–2006 Daniel Davison (PhD student, Comm Ev. Biol.; joint with S. Hackett)
now postdoc in Statistics with P. Donnelly, U. of Oxford
- 2001–2005 Sebastian Zöllner (postdoc); now assistant professor, Biostatistics, U. of Michigan
- 2002–2004 Giovanni Montana (postdoc); now junior faculty, Statistics, Imperial College London
- 2002–2003 Jeffrey Wall (postdoc); now assistant professor, UCSF

Software (<http://pritch.bsd.uchicago.edu>)

- structure* (2000) MCMC program for using multilocus genotype data to address various questions relating to population structure.
(2002) Version 2.0 released (updated and extended, with graphical front end)
- STRAT* (2000) Program for association mapping in structured populations.
- TreeLD* (2004) MCMC coalescent program for association mapping.
- MALDSoft* (2004) Program for admixture mapping.
- Haplotter* (2006) Web browser for scanning signals of selection in HapMap.

Downloads: ~1500 registered software downloads per year not including Haplotter (as of 2007).

Research Interests

My research group applies statistical or mathematical approaches to problems in human genetics and population/evolutionary genetics. We are interested both in developing new statistical methods for genetic analysis, and in performing statistical analyses of existing data from humans and other organisms. Much of our work makes use of computationally intensive approaches such as Markov chain Monte Carlo; these approaches can be effective for extracting subtle signals from large and complex data sets.

Current research areas include: (i) development of statistical methods for mapping complex trait genes; (ii) studies of linkage disequilibrium and other aspects of genetic variation to learn about natural selection and population history; (iii) use of genome-wide SNP data sets to study structural polymorphisms including copy number polymorphisms and inversions; (iv) inference of population structure from multilocus genetic data; (v) comparative genomics of non-coding DNA.

Past research contributions include:

- Inference of population structure. My colleagues and I have developed a model-based clustering approach for using multilocus genotype data to infer population structure, and assign individuals to populations [31, 19]. This method is now widely used in a range of fields—for instance: to study the structure of human populations [21]; to investigate the evolution of pathogens [20]; and in conservation genetics. Our original paper won an award from the International Society for Bayesian Applications, and our application to human data won Lancet's Paper of the Year award.
- 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. My work in this area has helped to show that these problems can be greatly reduced by using random unlinked marker loci to detect, and correct for cryptic population structure [32, 30, 23].
- Human variation and demographic history. I have a long-standing interest in using patterns of genetic variation to learn about evolutionary or historical processes. My 1999 paper on inferring human demographic history from Y chromosome data [33] was among the first papers to apply Approximate Bayesian Computation in genetics. Another focus of interest has been on how demography and the biology of recombination shape linkage disequilibrium in humans [26, 16, 3]. More recently, our focus has partly switched to studies of natural selection in humans, including a recent paper showing widespread signals of very strong selection in humans over the last $\sim 10,000$ years [5] as well recent work on the relationship between humans and Neanderthals [2].

A new line of research for us, supported by a Packard grant, deals with the population genetics of structural variation. Our first paper in this area shows that deletion polymorphism is extremely widespread in the human genome [6]. We are continuing this line of work in collaboration with Carole Ober in her genetic studies of the Hutterites.

- Population genetic models for gene mapping. A fourth area of interest is in applying coalescent modeling and inference techniques to complex trait mapping. I have used population genetic arguments to outline the scenarios under which the common disease-common variant hypothesis is, and is not, likely to hold [24, 22]. A more recent focus is on developing an MCMC-based coalescent approach to multipoint association mapping and fine mapping [10].

Publications (See <http://pritch.bsd.uchicago.edu> for additional lab publications)

- [1] 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. Gori, 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.
- [2] 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.
- [3] 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.
- [4] 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.
- [5] 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.
- [6] 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.
- [7] 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.
- [8] 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.
- [9] B. F. Voight and J. K. Pritchard. Confounding from cryptic relatedness in case-control association studies. *PLoS Genetics*, 1(e32):1–10, 2005.
- [10] S. Zöllner and J. K. Pritchard. Coalescent-based association mapping and fine mapping of complex trait loci. *Genetics*, 169:1071–1092, 2005.
- [11] S. Zollner, X. Wen, and J. K. Pritchard. Association mapping and fine mapping with TreeLD. *Bioinformatics*, 21:3168–70, 2005.
- [12] 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.
- [13] 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.
- [14] 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.
- [15] 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.
- [16] 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.

- [17] 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.
- [18] 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" [technical comment]. *Science*, 300:1877, 2003.
- [19] 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.
- [20] 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.
- [21] 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.
- [22] 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.
- [23] J. K. Pritchard and P. Donnelly. Case-control studies of association in structured or admixed populations. *Theor Popul Biol*, 60:227–37, 2001.
- [24] J. K. Pritchard. Are rare variants responsible for susceptibility to common diseases? *Am. J. Hum. Genet.*, 69:124–137, 2001.
- [25] J. K. Pritchard. Deconstructing maize population structure. *Nature Genetics*, 28:203–204, 2001.
- [26] J. K. Pritchard and M. Przeworski. Linkage disequilibrium in humans: models and data. *Am. J. Hum. Genet.*, 69:1–14, 2001.
- [27] 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.
- [28] R. Thompson, J. K. Pritchard, P. Shen, P. J. Oefner, and M. W. Feldman. Recent common ancestry of human Y chromosomes: Evidence from DNA sequence data. *Proc. Natl. Acad. Sci. USA*, 97:7360–7365, 2000.
- [29] 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.
- [30] J. K. Pritchard, M. Stephens, N. A. Rosenberg, and P. Donnelly. Association mapping in structured populations. *Am. J. Hum. Genet.*, 67:170–181, 2000.
- [31] J. K. Pritchard, M. Stephens, and P. Donnelly. Inference of population structure using multilocus genotype data. *Genetics*, 155:945–959, 2000.
- [32] 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.
- [33] 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.
- [34] 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.

- [35] 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.
- [36] 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.
- [37] C. T. Bergstrom and J. K. Pritchard. Germline bottlenecks and the evolutionary maintenance of mitochondrial genomes. *Genetics*, 149:2135–2146, 1998.
- [38] J. K. Pritchard and S. W. Schaeffer. Polymorphism and divergence at a *Drosophila melanogaster* pseudogene locus. *Genetics*, 147:199–208, 1997.
- [39] J. K. Pritchard and M. W. Feldman. Statistics for microsatellite variation based on coalescence. *Theor. Pop. Biol.*, 50:325–344, 1996.
- [40] J. K. Pritchard and M. W. Feldman. Genetic data and the African origin of modern humans [technical comment]. *Science*, 274:1548, 1996.
- [41] W. G. Pritchard and J. K. Pritchard. Mathematical models of running. *American Scientist*, 82:546–553, 1994.

Invited Talks Given

- 2007 Department of Genome Sciences, University of Washington
- 2006 American College of Neuropharmacology annual meeting (Plenary), Hollywood FL
 “Genome-wide association studies: design and analysis.”, Yale
 American Society Human Genetics (from submitted abstract; also session chair), New Orleans
 “Human Genome Variation 2006”, Hong Kong
 Genome Sciences Training Program Annual Retreat, University of Wisconsin
 “Biology of Genomes”, Cold Spring Harbor
 “Beyond HapMap”, Cambridge MA (from submitted abstract)
 “Chicago Convenes”, UC alumni lecture
 Program for Interdisc. Maths, Ecology, Stats; Colorado State University
 Conference: “Genome Sequence Variation”, Keystone Symposium
- 2005 “Third Seattle Symposium in Biostatistics” (teaching lectures)
 Field Museum Seminar Series, Chicago
 Department of Human Genetics, UCLA
 Department of Genetics (Annual Retreat), Stanford University
 Packard Foundation Meeting, Monterey
 Conference: “Evolutionary Genomics Workshop”, Mathematical Biosciences Institute, Columbus
 Conference: “Genomic Studies and the HapMap”, Oxford UK
 Department of Biostatistics, University of Alabama, Birmingham
 Department of Genetics, Case Western University
 Center for Human Genetics, University of Texas, Southwestern
- 2004 Takeda Foundation Symposium on Bioscience: “Genome Analysis and Medicine”, Tokyo
 10th Samsung International Symposium on Molecular Medicine, Seoul
 USC (CEGS) mini-symposium on population genetics of linkage disequilibrium, Los Angeles
 Mathematical Genetics Symposium, University of Durham UK
 Department of Human Genetics, University of Iowa
 Perlegen Sciences, Mountain View CA

Program in Bioinformatics, North Carolina State University (student-invited speaker)
 International Biometric Society: ENAR spring meeting, Pittsburgh
 NIMH conference: "Gene discovery in mental disorders", Laguna Beach CA (discussant)
 Department of Human Genetics, Emory University
 2003 Department of Genome Sciences, University of Washington
 6th International SNP Meeting, Virginia
 Henry Stewart Talks: Genetic Analysis in Drug R&D (talk series available for online purchase)
 American Association for Cancer Research: "SNPs, haplotypes and cancer", Key Biscayne FL
 Genetics of Complex Diseases and Isolated Populations, Sardinia Italy
 American Thoracic Society Conference (workshop on pop. gen. approaches) Seattle WA
 Marshfield Medical Research Foundation/Mammalian Genotyping Service, Marshfield WI
 Department of Biology, University of Illinois, Chicago
 Department of Statistics, University of Oxford
 Max Planck Institute for Evolutionary Biology, Leipzig Germany
 Testing of Spatial Structure Models Workshop (Scripps)
 2002 DIMACS (Rutgers) Workshop on computational methods for SNPs and haplotype inference
 Am. Soc. Hum Genet., slide presentation (from submitted abstract)
 5th International SNP Meeting, Reykjavik
 Conference: "Relating Genetic Variation to Disease Workshop", NHGRI
 Genetics group, Medical College of Wisconsin
 Banbury Meeting, "Sequence-based disease discovery", Cold Spring Harbor
 2001 Conference: "Genetic Analysis of Multifactorial Diseases", Tokyo
 Conference: "South and North of England Genetic Epidemiology" London
 Department of Ecology and Evolution, Purdue, IN
 Biostatistics Department, University of Michigan
 MCB and Genetics Department, Cornell University
 2000 Conference: "Genetics of Cardiovascular Disease workshop", London
 Royal Statistical Society Meeting, Reading, UK
 Conservation group, Universite Joseph Fourier, Grenoble
 Human Genetics Department, University of Chicago
 Genetics Department, University of Washington
 Genetics Department, University of Michigan
 1999 Mathematics Department, University of Essex, UK
 Biological Anthropology Department, University of Oxford
 Human Genetics Department, Washington University in St. Louis
 Statistics Department, University of Reading, UK
 Biology Department, University College London
 1997 Zoology Department, University of Oxford
 1995 Biostatistics Workshop, Stanford University