

Joseph K. Pickrell

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Employment History

2014-present Junior Group Leader and Core Member, New York Genome Center
2014-present Adjunct Assistant Professor, Department of Biological Sciences, Columbia University
2011-2013 Postdoctoral fellow, advised by David Reich
Department of Genetics, Harvard Medical School

Education

2011 PhD, advised by Jonathan Pritchard
Department of Human Genetics, University of Chicago
2006 Masters (M2) in Genetic Epidemiology, advised by Catherine Bourgain
Université de Paris-Sud
2005 B.S. in Biology, honors thesis advised by Todd Vision
University of North Carolina, Chapel Hill

Research Support

Current:
2015-2018 R01 MH106842: "Statistical models for fine-mapping and interpretation of schizophrenia-associated loci"
Completed:
2012-2013 F32 GM103098: "Population genetics methods for inference of ancestry and association mapping in diverse populations"

Research Interests

I use computational and statistical tools to address the question: what are the causes and consequences of genetic variation within and between human populations? I work to understand the evolutionary forces that act on genetic variation, as well as the molecular and phenotypic consequences of this variation. My research involves the development of statistical models for integrating diverse sources of data, with application to many areas of human genetics, including genomic approaches to understanding gene regulation, mapping genetic variants that influence traits, and inference about natural selection and demography.

Publications

[1] J. K. Pickrell, "Fulfilling the promise of Mendelian randomization," *bioRxiv*, 2015.

- [2] **J. K. Pickrell** and D. Reich, "Toward a new history and geography of human genes informed by ancient DNA," *Trends in Genetics*, vol. 30, no. 9, pp. 377–89, Sep 2014.
- [3] J. A. Hodgson, **J. K. Pickrell**, L. N. Pearson, E. E. Quillen, A. Prista, J. Rocha, H. Soodyall, M. D. Shriver, and G. H. Perry, "Natural selection for the Duffy-null allele in the recently admixed people of Madagascar," *Proc Biol Sci*, vol. 281, no. 1789, p. 20140930, Aug 2014.
- [4] B. Pasaniuc, N. Zaitlen, H. Shi, G. Bhatia, A. Gusev, **J. Pickrell**, J. Hirschhorn, D. P. Strachan, N. Patterson, and A. L. Price, "Fast and accurate imputation of summary statistics enhances evidence of functional enrichment," *Bioinformatics*, Jul 2014.
- [5] **J. K. Pickrell**, "Joint analysis of functional genomic data and genome-wide association studies of 18 human traits," *American Journal of Human Genetics*, vol. 94, no. 4, pp. 559–73, Apr 2014.
- [6] **J.K. Pickrell**, N. Patterson, P.-R. Loh, M. Lipson, B. Berger, M. Stoneking, B. Pakendorf, and D. Reich, "Ancient west Eurasian ancestry in southern and eastern Africa," *Proc Natl Acad Sci U S A*, vol. 111, no. 7, pp. 2632–7, Feb 2014.
- [7] K. Prüfer, F. Racimo, N. Patterson, F. Jay, S. Sankararaman, S. Sawyer, A. Heinze, G. Renaud, P. H. Sudmant, C. de Filippo, H. Li, S. Mallick, M. Dannemann, Q. Fu, M. Kircher, M. Kuhlwilm, M. Lachmann, M. Meyer, M. Ongyerth, M. Siebauer, C. Theunert, A. Tandon, P. Moorjani, **J. K. Pickrell**, J. C. Mullikin, S. H. Vohr, R. E. Green, I. Hellmann, P. L. F. Johnson, H. Blanche, H. Cann, J. O. Kitzman, J. Shendure, E. E. Eichler, E. S. Lein, T. E. Bakken, L. V. Golovanova, V. B. Doronichev, M. V. Shunkov, A. P. Derevianko, B. Viola, M. Slatkin, D. Reich, J. Kelso, and S. Pääbo, "The complete genome sequence of a Neanderthal from the Altai Mountains," *Nature*, vol. 505, no. 7481, pp. 43–9, Jan 2014.
- [8] G. Brandt, W. Haak, C. J. Adler, C. Roth, A. Szécsényi-Nagy, S. Karimnia, S. Möller-Rieker, H. Meller, R. Ganslmeier, S. Friederich, V. Dresely, N. Nicklisch, **J. K. Pickrell**, F. Sirocko, D. Reich, A. Cooper, K. W. Alt, and Genographic Consortium, "Ancient DNA reveals key stages in the formation of central European mitochondrial genetic diversity," *Science*, vol. 342, no. 6155, pp. 257–61, Oct 2013.
- [9] P.-R. Loh, M. Lipson, N. Patterson, P. Moorjani, **J. K. Pickrell**, D. Reich, and B. Berger, "Inferring admixture histories of human populations using linkage disequilibrium," *Genetics*, vol. 193, no. 4, pp. 1233–54, Apr 2013.
- [10] **J. K. Pickrell** and J. K. Pritchard, "Inference of population splits and mixtures from genome-wide allele frequency data," *PLoS Genetics*, vol. 8, no. 11, p. e1002967, Nov 2012.
- [11] **J. K. Pickrell**, N. Patterson, C. Barbieri, F. Berthold, L. Gerlach, T. Güldemann, B. Kure, S. W. Mpoloka, H. Nakagawa, C. Naumann, M. Lipson, P.-R. Loh, J. Lachance, J. Mountain, C. D. Bustamante, B. Berger, S. A. Tishkoff, B. M. Henn, M. Stoneking, D. Reich, and B. Pakendorf, "The genetic prehistory of southern Africa," *Nature Communications*, vol. 3, p. 1143, Oct 2012.
- [12] **J. K. Pickrell**, Y. Gilad, and J. K. Pritchard, "Comment on "Widespread RNA and DNA sequence differences in the human transcriptome"," *Science*, vol. 335, no. 6074, p. 1302, Mar 2012.
- [13] 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*, vol. 8, no. 10, p. e1003000, Oct 2012.
- [14] L. Jostins, **J. K. Pickrell**, D. G. MacArthur, and J. C. Barrett, "Misuse of hierarchical linear models overstates the significance of a reported association between OXTR and prosociality," *PNAS*, vol. 109, no. 18, p. E1048, May 2012.

- [15] 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*, vol. 7, no. 2, p. e30629, 2012.
- [16] D. G. MacArthur, S. Balasubramanian, A. Frankish, N. Huang, J. Morris, K. Walter, L. Jostins, L. Habegger, **J. K. Pickrell**, S. B. Montgomery, C. A. Albers, Z. D. Zhang, D. F. Conrad, G. Lunter, H. Zheng, Q. Ayub, M. A. DePristo, E. Banks, M. Hu, R. E. Handsaker, J. A. Rosenfeld, M. Fromer, M. Jin, X. J. Mu, E. Khurana, K. Ye, M. Kay, G. I. Saunders, M.-M. Suner, T. Hunt, I. H. A. Barnes, C. Amid, D. R. Carvalho-Silva, A. H. Bignell, C. Snow, B. Yngvadottir, S. Bumpstead, D. N. Cooper, Y. Xue, I. G. Romero, 1000 Genomes Project Consortium, J. Wang, Y. Li, R. A. Gibbs, S. A. McCarroll, E. T. Dermitzakis, J. K. Pritchard, J. C. Barrett, J. Harrow, M. E. Hurles, M. B. Gerstein, and C. Tyler-Smith, "A systematic survey of loss-of-function variants in human protein-coding genes," *Science*, vol. 335, no. 6070, pp. 823–8, Feb 2012.
- [17] 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, "DNase I sensitivity QTLs are a major determinant of human expression variation," *Nature*, vol. 482, no. 7385, pp. 390–4, Feb 2012.
- [18] 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*, vol. 22, no. 4, pp. 602–10, Apr 2012.
- [19] **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*, vol. 27, no. 15, pp. 2144–6, Aug 2011 [*co-first authors].
- [20] 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*, vol. 12, no. 1, p. R10, 2011.
- [21] **J. K. Pickrell**, A. A. Pai, Y. Gilad, and J. K. Pritchard, "Noisy splicing drives mRNA isoform diversity in human cells," *PLoS Genetics*, vol. 6, no. 12, p. e1001236, 2010.
- [22] G. Kosova, **J. K. Pickrell**, J. L. Kelley, P. F. McArdle, A. R. Shuldiner, M. Abney, and C. Ober, "The CFTR Met 470 allele is associated with lower birth rates in fertile men from a population isolate," *PLoS Genetics*, vol. 6, no. 6, p. e1000974, Jun 2010.
- [23] **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*, vol. 464, no. 7289, pp. 768–72, Apr 2010.
- [24] J. K. Pritchard, **J. K. Pickrell**, and G. Coop, "The genetics of human adaptation: hard sweeps, soft sweeps, and polygenic adaptation," *Current Biology*, vol. 20, no. 4, pp. R208–15, Feb 2010.
- [25] G. H. Perry and **J. K. Pickrell**, "A rod cell marker of nocturnal ancestry [News and Views]," *Journal of Human Evolution*, vol. 58, no. 2, pp. 207–10, Feb 2010.
- [26] 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*, vol. 25, no. 24, pp. 3207–12, Dec 2009.
- [27] L. B. Barreiro, M. Ben-Ali, H. Quach, G. Laval, E. Patin, **J. K. Pickrell**, C. Bouchier, M. Tichit, O. Neyrolles, B. Gicquel, J. R. Kidd, K. K. Kidd, A. Alcaïs, J. Ragimbeau, S. Pellegrini, L. Abel, J.-L. Casanova, and L. Quintana-Murci, "Evolutionary dynamics of human Toll-like receptors and their different contributions to host defense," *PLoS Genetics*, vol. 5, no. 7, p. e1000562, Jul 2009.

- [28] D. A. Faddah, E. W. Ganko, C. McCoach, **J. K. Pickrell**, S. E. Hanlon, F. G. Mann, J. O. Mieczkowska, C. D. Jones, J. D. Lieb, and T. J. Vision, "Systematic identification of balanced transposition polymorphisms in *Saccharomyces cerevisiae*," *PLoS Genetics*, vol. 5, no. 6, p. e1000502, Jun 2009.
- [29] 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*, vol. 5, no. 6, p. e1000500, Jun 2009 [*co-first authors].
- [30] **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*, vol. 19, no. 5, pp. 826–37, May 2009.
- [31] C. Dalmasso, **J. K. Pickrell**, M. Tuefferd, E. Génin, C. Bourgain, and P. Broët, "A mixture model approach to multiple testing for the genetic analysis of gene expression," *BMC Proceedings*, vol. 1 Suppl 1, p. S141, 2007.
- [32] **J. K. Pickrell**, F. Clerget-Darpoux, and C. Bourgain, "Power of genome-wide association studies in the presence of interacting loci," *Genetic Epidemiology*, vol. 31, no. 7, pp. 748–62, Nov 2007.
- [33] S. I. Sharp, **J. K. Pickrell**, and C. L. Jahn, "Identification of a novel "chromosome scaffold" protein that associates with Tec elements undergoing en masse elimination in *Euplotes crassus*," *Molecular Biology of the Cell*, vol. 14, no. 2, pp. 571–84, Feb 2003.

Awards and Honors

2013	Named to the 2013 Young Investigators list by GenomeWeb
2012-2013	NIH postdoctoral fellowship (NRSA)
2012	Outstanding thesis award, University of Chicago
2009	Trainee Research Award, American Society for Human Genetics
2006-2009	Genetics and Regulation Training Grant
2005	Graduated with highest honors from UNC
2001-2005	National Merit Scholar

Teaching Experience

2013	Instructor, Montreal Spring School of Population Genomics
2012	Instructor, Montreal Spring School of Population Genomics
2011	Tutor, Okinawa Institute of Science and Technology course on Quantitative Evolutionary and Comparative Genomics
2008	Teaching Assistant, University of Chicago <i>Genetics, Ancestry, and Medicine</i> [undergraduate]
2007	Teaching Assistant, University of Chicago <i>Statistical Genetics I</i> [graduate]

Invited talks

- 2015** The Future of Genomic Medicine, La Jolla, CA
Center for Bioinformatics and Computational Biology, University of Maryland
- 2014** Finnish Institute for Molecular Medicine, Helsinki, Finland
Penn Bioinformatics Forum, University of Pennsylvania, Philadelphia, PA
Institute for Computational Biomedicine, Weill Cornell Medical College, New York, NY
Department of Biology, Brooklyn College, Brooklyn, NY
The Biology of Genomes [from abstract], Cold Spring Harbor, NY
Sanger Institute, Cambridge, UK
Biological Sequence Analysis Workshop, Oxford, UK
Joint Statistical Meetings, Boston, MA
- 2013** Department of Biology, NYU, New York, NY
Department of Statistics, Columbia University, New York, NY
Center for Computational Biology, University of California, Berkeley, CA
Sanger Institute, Cambridge, UK
The Biology of Genomes [from abstract], Cold Spring Harbor, NY
Montreal Spring School of Population Genomics, Montreal, Quebec
Institute for Human Genetics, UCSF, San Francisco, CA
Society for Molecular Biology and Evolution [from abstract], Chicago, IL
American Society for Human Genetics [from abstract], Boston, MA
Adelaide Center for Ancient DNA, Adelaide, Australia
Department of Human Genetics, UCLA, Los Angeles, CA
- 2012** Synbreed School in Population Genomics, Salzburg, Austria
American Society for Human Genetics, San Francisco, CA
Montreal Spring School of Population Genomics, Montreal, Quebec
Bauer Forum, Harvard University, Cambridge, MA
- 2011** Department of Computer Science, MIT, Cambridge, MA
Department of Genetics, Stanford University, Palo Alto, CA
Okinawa Institute of Science and Technology, Okinawa, Japan
- 2010** Newton Institute, Cambridge, UK
Department of Statistics, Oxford University, Oxford, UK
- 2009** Primate Research Institute, Kyoto University, Inuyama, Japan
- 2008** Society for Molecular Biology and Evolution [from abstract], Barcelona, Spain
Pasteur Institute, Paris, France
- 2007** European Society for Human Genetics [from abstract], Amsterdam, Netherlands