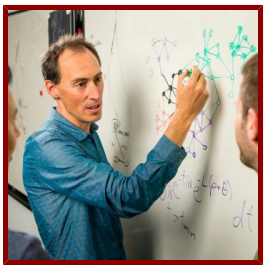
Stanford



Jonathan Pritchard

Bing Professor of Population Studies, Professor of Genetics and Biology

Bio

BIO

Jonathan Pritchard is the Bing Professor of Population Studies, in the departments of Genetics and Biology at Stanford University. He grew up in England, and studied at Penn State, Stanford, and Oxford. He joined the faculty of the University of Chicago in 2001 and returned to Stanford University in 2013. His lab has done wide-ranging research on using genetics to study human population structure, history, and adaptation, and on understanding the mechanisms by which genetic variation affects gene regulation and complex traits. One of his early contributions was the Structure algorithm for using genetic data to infer population structure and personal ancestry, which has been widely influential and cited more than 40,000 times. More recently, his lab has made essential contributions on modeling and interpreting the genetic basis of complex traits and diseases. A current focus is on how to use experimental perturbation methods to model human gene regulatory networks for understanding complex traits, with a particular focus on the immune system. He has been recognized with various honors, including election to the National Academy of Sciences and the American Academy of Arts and Sciences.

ACADEMIC APPOINTMENTS

- Professor, Genetics
- Professor, Biology
- Member, Bio-X

ADMINISTRATIVE APPOINTMENTS

- Co-Director, Stanford's Center for Computational, Evolutionary and Human Genomics, (2017- present)
- Investigator, Howard Hughes Medical Institute, (2008-2019)
- Professor, University of Chicago (Human Genetics), (2006-2013)
- Assistant Professor, University of Chicago (Human Genetics), (2001-2005)
- Postdoc, University of Oxford (Statistics), (1998-2001)

HONORS AND AWARDS

- Fellow, National Academy of Sciences (2025)
- Appointed Bing Professor of Population Studies, Stanford University (2020)
- Fabio Frassetto International Prize in Physical Anthropology, Lincean Academy of Italy (2019)
- Kistler Prize in Population Genetics and Society, Stanford University (2015)
- Fellow, American Academy of Arts and Sciences (2013)
- Novitski Prize, Genetics Society of America (2013)

- Investigator, Howard Hughes Medical Institute (2008-2019)
- Packard Fellow, Packard Foundation (2004)
- Sloan Fellow, Sloan Foundation (2004)
- Mitchell Prize, American Statistical Association and the International Society of Bayesian Analysis (2002)
- Paper of the Year (Rosenberg et al 2002), Lancet (2002)

PROFESSIONAL EDUCATION

- Ph.D., Stanford University , Biology (1998)
- B.Sc., Pennsylvania State University , Biology and Mathematics (1994)

LINKS

- Lab web site: http://pritchardlab.stanford.edu

Research & Scholarship

CURRENT RESEARCH AND SCHOLARLY INTERESTS

My group has expertise in the development of new statistical methods for genetic analysis and in their application to genomic data from humans and other organisms. We focus on questions relating to genetic variation and evolution: How does genetic variation impact phenotypic traits and evolution, both at the organismal and cellular level? What can we learn from genome sequences of modern and ancient humans about the relationships among human populations, and the the nature of adaptation in these populations?

We often work on problems where there are no off-the-shelf statistical methods. Thus, an important part of our work is in developing appropriate statistical and computational approaches that can yield new insights into biological data. In the past, we have made important contributions to a variety of problems in human population genetics, including methods for complex trait mapping, inference of population structure and history, and studies of natural selection. We have a strong track record of producing user-friendly resources that are widely used in the community, and in applied data analysis to tackle important biological questions. Notably, our Structure algorithm and software package for inferring population structure from genetic data have received >30,000 total citations spread across several papers.

Since 2008 an important emphasis of my group has focused on understanding gene regulation, and in particular how genetic variation may impact regulation. Ultimately, we would like to be able to predict which noncoding variants in the genome are likely to have regulatory effects in any given cell type, and how these link to phenotypic variation and disease. My lab has been deeply involved in developing new computational methods to interpret various types of modern genomic assays and in linking these to genetic variation.

Secondly, we have had a major focus on understanding the genetic architecture of complex traits, and the implications for understanding evolution. We have argued that much--if not most--evolution in humans likely proceeds through a process that we call "polygenic adaptation" in which populations evolve through small allele frequency shifts at many loci.

We have also written extensively about conceptual models for understanding the genetic architecture of trait variation (Boyle et al, 2017). We have argued that the data are consistent with a model in which essentially every regulatory variant in disease-relevant cell types can affect risk, and proposed that most of these effects act through trans-regulatory networks. Testing this model is an ongoing focus of our work.

Teaching

COURSES

2026-27

- Advanced Genetics: GENE 205 (Win)
- Biology PhD Lab Rotation: BIO 299 (Aut, Win, Spr)
- Culture, Evolution, and Society: HUMBIO 2B (Aut)
- Genomic approaches to the study of human disease: BIO 127, BIO 247, GENE 247 (Win)

2025-26

- Advanced Genetics: GENE 205 (Win)
- Culture, Evolution, and Society: HUMBIO 2B (Aut)

2024-25

- Advanced Genetics: GENE 205 (Win)
- Biology PhD Lab Rotation: BIO 299 (Aut, Win)
- Culture, Evolution, and Society: HUMBIO 2B (Aut)
- Genomic approaches to the study of human disease: BIO 127, BIO 247, GENE 247 (Win)

2023-24

- Advanced Genetics: GENE 205 (Win)
- Biology PhD Lab Rotation: BIO 299 (Aut, Win, Spr)
- Culture, Evolution, and Society: HUMBIO 2B (Aut)
- Genomic approaches to the study of human disease: BIO 127, BIO 247, GENE 247 (Win)

STANFORD ADVISEES

Doctoral Dissertation Reader (AC)

Connor Duffy, Keala Gapin, Peter Gerlach, Ilayda Ilerten, Egor Lappo, Gabe Preising, Jess Rhodes

Postdoctoral Faculty Sponsor

Emma Dann, Yun Deng, Xinyi Li, Wangjie Liu, Jiacheng Miao, Leqi Xu

Doctoral Dissertation Advisor (AC)

Tami Gjorgjieva, Nikhil Milind, Courtney Smith, Tony Zeng, Julie Zhu

Doctoral Dissertation Co-Advisor (AC)

Alvina Adimoelja, Ryan Goto

Undergraduate Major Advisor

Mason Chen, Julieta Lamm-Perez, Jacob Shafik

GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Biology (School of Humanities and Sciences) (PhD Program)
- Biomedical Data Science (PhD Program)
- Genetics (PhD Program)

Publications

PUBLICATIONS

- **A way to identify the biological basis of gene-trait associations** *NATURE*
  Ota, M., Pritchard, J. K.
  2026

- **Causal modelling of gene effects from regulators to programs to traits.** *Nature*
  Ota, M., Spence, J. P., Zeng, T., Dann, E., Milind, N., Marson, A., Pritchard, J. K.
  2025

- **The primate Major Histocompatibility Complex as a case study of gene family evolution.** *eLife*
  Fortier, A. L., Pritchard, J. K.
  2025; 14

- **The primate Major Histocompatibility Complex as a case study of gene family evolution** *ELIFE*
  Fortier, A., Pritchard, J. K.
  2025; 14

- **Specificity, length and luck drive gene rankings in association studies.** *Nature*
  Spence, J. P., Mostafavi, H., Ota, M., Milind, N., Gjorgjieva, T., Smith, C. J., Simons, Y. B., Sella, G., Pritchard, J. K.
  2025

- **Simple scaling laws control the genetic architectures of human complex traits.** *PLoS biology*
  Simons, Y. B., Mostafavi, H., Zhu, H., Smith, C. J., Pritchard, J. K., Sella, G.
  2025; 23 (10): e3003402

- **Ancient trans-species polymorphism at the Major Histocompatibility Complex in primates.** *eLife*
  Fortier, A. L., Pritchard, J. K.
  2025; 14

- **rRNA paralogs with variations, rRNA-subtypes, affect diverse human phenotypes.** *medRxiv : the preprint server for health sciences*
  Rothschild, D., Pritchard, J. K., Barna, M.
  2025

- **Gene regulatory network structure informs the distribution of perturbation effects.** *PLoS computational biology*
  Aguirre, M., Spence, J. P., Sella, G., Pritchard, J. K.
  2025; 21 (9): e1013387

- **Buffering and non-monotonic behavior of gene dosage response curves for human complex traits.** *medRxiv : the preprint server for health sciences*
  Milind, N., Smith, C. J., Zhu, H., Gjorgjieva, T., Spence, J. P., Pritchard, J. K.
  2025

- **Regulatory network topology and the genetic architecture of gene expression.** *bioRxiv : the preprint server for biology*
  Aguirre, M., Spence, J. P., Sella, G., Pritchard, J. K.
  2025

- **Investigating the Role of Neighborhood Socioeconomic Status and Germline Genetics on Prostate Cancer Risk.** *HGG advances*
  Judd, J., Spence, J. P., Pritchard, J. K., Kachuri, L., Witte, J. S.
  2025: 100492

- **Haplotype analysis reveals pleiotropic disease associations in the HLA region.** *American journal of human genetics*
  Smith, C. J., Strausz, S., Spence, J. P., Ollila, H. M., Pritchard, J. K.
  2025

- **Transfer learning reveals sequence determinants of the quantitative response to transcription factor dosage.** *Cell genomics*
  Naqvi, S., Kim, S., Tabatabaee, S., Pampari, A., Kundaje, A., Pritchard, J. K., Wysocka, J.

2025: 100780

- **Causal modeling of gene effects from regulators to programs to traits: integration of genetic associations and Perturb-seq.** *bioRxiv : the preprint server for biology*
  Ota, M., Spence, J. P., Zeng, T., Dann, E., Marson, A., Pritchard, J. K.
  2025
- **Characterizing selection on complex traits through conditional frequency spectra.** *Genetics*
  Patel, R. A., WeiSS, C. L., Zhu, H., Mostafavi, H., Simons, Y. B., Spence, J. P., Pritchard, J. K.
  2024
- **Specificity, length, and luck: How genes are prioritized by rare and common variant association studies.** *bioRxiv : the preprint server for biology*
  Spence, J. P., Mostafavi, H., Ota, M., Milind, N., Gjorgjieva, T., Smith, C. J., Simons, Y. B., Sella, G., Pritchard, J. K.
  2024
- **Central control of dynamic gene circuits governs T cell rest and activation.** *Nature*
  Arce, M. M., Umhoefer, J. M., Arang, N., Kasinathan, S., Freimer, J. W., Steinhart, Z., Shen, H., Pham, M. T., Ota, M., Wadhera, A., Dajani, R., Dorovskyi, D., Chen, et al
  2024
- **Gene regulatory network inference from CRISPR perturbations in primary CD4+ T cells elucidates the genomic basis of immune disease.** *Cell genomics*
  Weinstock, J. S., Arce, M. M., Freimer, J. W., Ota, M., Marson, A., Battle, A., Pritchard, J. K.
  2024: 100671
- **The Primate Major Histocompatibility Complex: An Illustrative Example of Gene Family Evolution.** *bioRxiv : the preprint server for biology*
  Lyn Fortier, A., Pritchard, J. K.
  2024
- **Deciphering the impact of genomic variation on function.** *Nature*
  2024; 633 (8028): 47-57
- **Diversity of ribosomes at the level of rRNA variation associated with human health and disease.** *Cell genomics*
  Rothschild, D., Susanto, T. T., Sui, X., Spence, J. P., Rangan, R., Genuth, N. R., Sinnott-Armstrong, N., Wang, X., Pritchard, J. K., Barna, M.
  2024: 100629
- **Investigating the Role of Neighborhood Socioeconomic Status and Germline Genetics on Prostate Cancer Risk.** *medRxiv : the preprint server for health sciences*
  Judd, J., Spence, J. P., Pritchard, J. K., Kachuri, L., Witte, J. S.
  2024
- **Haplotype Analysis Reveals Pleiotropic Disease Associations in the HLA Region.** *medRxiv : the preprint server for health sciences*
  Smith, C. J., Strausz, S., Spence, J. P., Ollila, H. M., Pritchard, J. K.
  2024
- **Genetic variants affect diurnal glucose levels throughout the day.** *bioRxiv : the preprint server for biology*
  Sinnott-Armstrong, N., Strausz, S., Urpa, L., Abner, E., Valliere, J., Palta, P., Dashti, H. S., Daly, M., Pritchard, J. K., Saxena, R., Jones, S. E., Ollila, H. M.
  2024
- **Bayesian estimation of gene constraint from an evolutionary model with gene features.** *Nature genetics*
  Zeng, T., Spence, J. P., Mostafavi, H., Pritchard, J. K.
  2024
- **Gene regulatory network structure informs the distribution of perturbation effects.** *bioRxiv : the preprint server for biology*
  Aguirre, M., Spence, J. P., Sella, G., Pritchard, J. K.
  2024
- **A model for accurate quantification of CRISPR effects in pooled FACS screens.** *bioRxiv : the preprint server for biology*
  Pimentel, H., Freimer, J. W., Arce, M. M., Garrido, C. M., Marson, A., Pritchard, J. K.

2024

- **Conditional frequency spectra as a tool for studying selection on complex traits in biobanks.** *bioRxiv : the preprint server for biology*
  Patel, R. A., Weiß, C. L., Zhu, H., Mostafavi, H., Simons, Y. B., Spence, J. P., Pritchard, J. K.
  2024
- **Transfer learning reveals sequence determinants of the quantitative response to transcription factor dosage.** *bioRxiv : the preprint server for biology*
  Naqvi, S., Kim, S., Tabatabaee, S., Pampari, A., Kundaje, A., Pritchard, J. K., Wysocka, J.
  2024
- **Ancient DNA in Context: Port Cities and Mobility in the Iron Age and Roman Mediterranean**
  Moots, H., Antonio, M., Gao, Z., Lucci, M., Bondioli, L., Sperduti, A., Candilio, F., Coppa, A., Pinhasi, R., Pritchard, J.
  WILEY.2024: 121
- **Stable population structure in Europe since the Iron Age, despite high mobility.** *eLife*
  Antonio, M. L., WeiSS, C. L., Gao, Z., Sawyer, S., Oberreiter, V., Moots, H. M., Spence, J. P., Cheronet, O., Zagorc, B., Praxmarer, E., Ozdogan, K. T., Demetz, L., Gelabert, et al
  2024; 13
- **Toward the Identifiability of Comparative Deep Generative Models**
  Lopez, R., Huetter, J., Hajiramezanali, E., Pritchard, J. K., Regev, A.
  edited by Locatello, F., Didelez
  JMLR-JOURNAL MACHINE LEARNING RESEARCH.2024: 868-912
- **Base-editing mutagenesis maps alleles to tune human T cell functions.** *Nature*
  Schmidt, R., Ward, C. C., Dajani, R., Armour-Garb, Z., Ota, M., Allain, V., Hernandez, R., Layeghi, M., Xing, G., Goudy, L., Dorovskyi, D., Wang, C., Chen, et al
  2023
- **Systematic differences in discovery of genetic effects on gene expression and complex traits.** *Nature genetics*
  Mostafavi, H., Spence, J. P., Naqvi, S., Pritchard, J. K.
  2023
- **The functional impact of rare variation across the regulatory cascade.** *Cell genomics*
  Li, T., Ferraro, N., Strober, B. J., Aguet, F., Kasela, S., Arvanitis, M., Ni, B., Wiel, L., Hershberg, E., Ardlie, K., Arking, D. E., Beer, R. L., Brody, et al
  2023; 3 (10): 100401
- **Scaling the Discrete-time Wright-Fisher Model to Biobank-scale Datasets.** *Genetics*
  Spence, J. P., Zeng, T., Mostafavi, H., Pritchard, J. K.
  2023
- **Gene regulatory network inference from CRISPR perturbations in primary CD4+ T cells elucidates the genomic basis of immune disease.** *bioRxiv : the preprint server for biology*
  Weinstock, J. S., Arce, M. M., Freimer, J. W., Ota, M., Marson, A., Battle, A., Pritchard, J. K.
  2023
- **CRISPR screens decode cancer cell pathways that trigger γδ T cell detection.** *Nature*
  Mamedov, M. R., Vedova, S., Freimer, J. W., Sahu, A. D., Ramesh, A., Arce, M. M., Meringa, A. D., Ota, M., Chen, P. A., Hanspers, K., Nguyen, V. Q., Takeshima, K. A., Rios, et al
  2023
- **A genetic history of continuity and mobility in the Iron Age central Mediterranean.** *Nature ecology & evolution*
  Moots, H. M., Antonio, M., Sawyer, S., Spence, J. P., Oberreiter, V., WeiSS, C. L., Lucci, M., Cherifi, Y. M., La Pastina, F., Genchi, F., Praxmeier, E., Zagorc, B., Cheronet, et al
  2023
- **A genome-wide genetic screen uncovers determinants of human pigmentation.** *Science (New York, N.Y.)*
  Bajpai, V. K., Swigut, T., Mohammed, J., Naqvi, S., Arreola, M., Tycko, J., Kim, T. C., Pritchard, J. K., Bassik, M. C., Wysocka, J.
  2023; 381 (6658): eade6289

- **On the number of genealogical ancestors tracing to the source groups of an admixed population.** *Genetics*
  Mooney, J. A., Agranat-Tamir, L., Pritchard, J. K., Rosenberg, N. A.
  2023; 224 (3)
- **Bayesian estimation of gene constraint from an evolutionary model with gene features.** *Research square*
  Zeng, T., Spence, J. P., Mostafavi, H., Pritchard, J. K.
  2023
- **Subfunctionalized expression drives evolutionary retention of ribosomal protein paralogs Rps27 and Rps27l in vertebrates.** *eLife*
  Xu, A. F., Molinuevo, R., Fazzari, E., Tom, H., Zhang, Z., Menendez, J., Casey, K. M., Ruggero, D., Hinck, L., Pritchard, J. K., Barna, M.
  2023; 12
- **Scaling the Discrete-time Wright Fisher model to biobank-scale datasets.** *bioRxiv : the preprint server for biology*
  Spence, J. P., Zeng, T., Mostafavi, H., Pritchard, J. K.
  2023
- **Bayesian estimation of gene constraint from an evolutionary model with gene features.** *bioRxiv : the preprint server for biology*
  Zeng, T., Spence, J. P., Mostafavi, H., Pritchard, J. K.
  2023
- **Narcolepsy risk loci outline role of T cell autoimmunity and infectious triggers in narcolepsy.** *Nature communications*
  Ollila, H. M., Sharon, E., Lin, L., Sinnott-Armstrong, N., Ambati, A., Yogeshwar, S. M., Hillary, R. P., Jolanki, O., Faraco, J., Einen, M., Luo, G., Zhang, J., Han, et al
  2023; 14 (1): 2709
- **A novel quantitative trait locus implicates Msh3 in the propensity for genome-wide short tandem repeat expansions in mice.** *Genome research*
  Maksimov, M. O., Wu, C., Ashbrook, D. G., Villani, F., Colonna, V., Mousavi, N., Ma, N., Lu, L., Pritchard, J. K., Goren, A., Williams, R. W., Palmer, A. A., Gymrek, et al
  2023
- **Precise modulation of transcription factor levels identifies features underlying dosage sensitivity.** *Nature genetics*
  Naqvi, S., Kim, S., Hoskens, H., Matthews, H. S., Spritz, R. A., Klein, O. D., Hallgrimsson, B., Swigut, T., Claes, P., Pritchard, J. K., Wysocka, J.
  2023
- **A comprehensive rRNA variation atlas in health and disease.** *bioRxiv : the preprint server for biology*
  Rothschild, D., Susanto, T. T., Spence, J. P., Genuth, N. R., Sinnott-Armstrong, N., Pritchard, J. K., Barna, M.
  2023
- **Learning Causal Representations of Single Cells via Sparse Mechanism Shift Modeling**
  Lopez, R., Tagasovska, N., Ra, S., Cho, K., Pritchard, J. K., Regev, A.
  edited by VanDerSchaar, M., Zhang, C., Janzing, D.
  JMLR-JOURNAL MACHINE LEARNING RESEARCH.2023: 662-691
- **Integrative analysis of metabolite GWAS illuminates the molecular basis of pleiotropy and genetic correlation.** *eLife*
  Smith, C. J., Sinnott-Armstrong, N., Cichonska, A., Julkunen, H., Fauman, E. B., Wurtz, P., Pritchard, J. K.
  2022; 11
- **RNA editing underlies genetic risk of common inflammatory diseases.** *Nature*
  Li, Q., Gloudemans, M. J., Geisinger, J. M., Fan, B., Aguet, F., Sun, T., Ramaswami, G., Li, Y. I., Ma, J. B., Pritchard, J. K., Montgomery, S. B., Li, J. B.
  2022
- **Systematic discovery and perturbation of regulatory genes in human T cells reveals the architecture of immune networks.** *Nature genetics*
  Freimer, J. W., Shaked, O., Naqvi, S., Sinnott-Armstrong, N., Kathiria, A., Garrido, C. M., Chen, A. F., Cortez, J. T., Greenleaf, W. J., Pritchard, J. K., Marson, A.
  2022
- **Genetic interactions drive heterogeneity in causal variant effect sizes for gene expression and complex traits.** *American journal of human genetics*

Patel, R. A., Musharoff, S. A., Spence, J. P., Pimentel, H., Tcheandjieu, C., Mostafavi, H., Sinnott-Armstrong, N., Clarke, S. L., Smith, C. J., V.A. Million Veteran Program,,, Durda, P. P., Taylor, K. D., et al

2022

- **A natural mutator allele shapes mutation spectrum variation in mice.** *Nature*

  Sasani, T. A., Ashbrook, D. G., Beichman, A. C., Lu, L., Palmer, A. A., Williams, R. W., Pritchard, J. K., Harris, K.

  2022

- **Author Correction: Perspectives on ENCODE.** *Nature*

  ENCODE Project Consortium, Snyder, M. P., Gingeras, T. R., Moore, J. E., Weng, Z., Gerstein, M. B., Ren, B., Hardison, R. C., Stamatoyannopoulos, J. A., Graveley, B. R., Feingold, E. A., Pazin, M. J., Pagan, M., et al

  2022

- **Author Correction: Expanded encyclopaedias of DNA elements in the human and mouse genomes.** *Nature*

  ENCODE Project Consortium, Moore, J. E., Purcaro, M. J., Pratt, H. E., Epstein, C. B., Shoresh, N., Adrian, J., Kawli, T., Davis, C. A., Dobin, A., Kaul, R., Halow, J., Van Nostrand, E. L., et al

  2022

- **Author Correction: Genetics of 35 blood and urine biomarkers in the UK Biobank.** *Nature genetics*

  Sinnott-Armstrong, N., Tanigawa, Y., Amar, D., Mars, N., Benner, C., Aguirre, M., Venkataraman, G. R., Wainberg, M., Ollila, H. M., Kiiskinen, T., Havulinna, A. S., Pirruccello, J. P., Qian, et al

  2021

- **Large-scale cis- and trans-eQTL analyses identify thousands of genetic loci and polygenic scores that regulate blood gene expression.** *Nature genetics*

  Vosa, U., Claringbould, A., Westra, H., Bonder, M. J., Deelen, P., Zeng, B., Kirsten, H., Saha, A., Kreuzhuber, R., Yazar, S., Brugge, H., Oelen, R., de Vries, et al

  2021

- **Shared heritability of human face and brain shape.** *Nature genetics*

  Naqvi, S., Sleyp, Y., Hoskens, H., Indencleef, K., Spence, J. P., Bruffaerts, R., Radwan, A., Eller, R. J., Richmond, S., Shriver, M. D., Shaffer, J. R., Weinberg, S. M., Walsh, et al

  2021

- **GWAS of three molecular traits highlights core genes and pathways alongside a highly polygenic background.** *eLife*

  Sinnott-Armstrong, N., Naqvi, S., Rivas, M., Pritchard, J. K.

  2021; 10

- **A regulatory variant at 3q21.1 confers an increased pleiotropic risk for hyperglycemia and altered bone mineral density.** *Cell metabolism*

  Sinnott-Armstrong, N., Sousa, I. S., Laber, S., Rendina-Ruedy, E., Nitter Dankel, S. E., Ferreira, T., Mellgren, G., Karasik, D., Rivas, M., Pritchard, J., Guntur, A. R., Cox, R. D., Lindgren, et al

  2021

- **Genetics of 35 blood and urine biomarkers in the UK Biobank.** *Nature genetics*

  Sinnott-Armstrong, N., Tanigawa, Y., Amar, D., Mars, N., Benner, C., Aguirre, M., Venkataraman, G. R., Wainberg, M., Ollila, H. M., Kiiskinen, T., Havulinna, A. S., Pirruccello, J. P., Qian, et al

  2021

- **Using CRISPR screens to map gene regulatory networks in primary human T cells**

  Shaked, O., Freimer, J., Pritchard, J., Marson, A.

  WILEY.2021: 41

- **Perspectives on ENCODE.** *Nature*

  ENCODE Project Consortium, Snyder, M. P., Gingeras, T. R., Moore, J. E., Weng, Z., Gerstein, M. B., Ren, B., Hardison, R. C., Stamatoyannopoulos, J. A., Graveley, B. R., Feingold, E. A., Pazin, M. J., Pagan, M., et al

  2020; 583 (7818): 693–98

- **The Genetics of Malaria Resistance in Ancient Rome**

  Moots, H., Pickel, D., Sperduti, A., Antonio, M. L., Gao, Z., Nava, A., Gelabert, P., Lucci, M., Candilio, F., Sawyer, S., Oberreiter, V., Rubini, M., Bondioli, et al

  WILEY.2020: 191–92

- **Variable prediction accuracy of polygenic scores within an ancestry group.** *eLife*
  Mostafavi, H., Harpak, A., Agarwal, I., Conley, D., Pritchard, J. K., Przeworski, M.
  2020; 9

- **Evolutionary Persistence of DNA Methylation for Millions of Years after Ancient Loss of a De Novo Methyltransferase.** *Cell*
  Catania, S., Dumesic, P. A., Pimentel, H., Nasif, A., Stoddard, C. I., Burke, J. E., Diedrich, J. K., Cook, S., Shea, T., Geinger, E., Lintner, R., Yates, J. R., Hajkova, et al
  2020

- **Chromatin accessibility dynamics in a model of human forebrain development.** *Science (New York, N.Y.)*
  Trevino, A. E., Sinnott-Armstrong, N. n., Andersen, J. n., Yoon, S. J., Huber, N. n., Pritchard, J. K., Chang, H. Y., Greenleaf, W. J., Pașca, S. P.
  2020; 367 (6476)

- **Expanded encyclopaedias of DNA elements in the human and mouse genomes.** *Nature*
  Moore, J. E., Purcaro, M. J., Pratt, H. E., Epstein, C. B., Shoresh, N. n., Adrian, J. n., Kawli, T. n., Davis, C. A., Dobin, A. n., Kaul, R. n., Halow, J. n., Van Nostrand, E. L., Freese, et al
  2020; 583 (7818): 699–710

- **Landscape of stimulation-responsive chromatin across diverse human immune cells.** *Nature genetics*
  Calderon, D., Nguyen, M. L., Mezger, A., Kathiria, A., Muller, F., Nguyen, V., Lescano, N., Wu, B., Trombetta, J., Ribado, J. V., Knowles, D. A., Gao, Z., Blaeschke, et al
  2019

- **Public Discussion Affects Question Asking at Academic Conferences.** *American journal of human genetics*
  Telis, N., Glassberg, E. C., Pritchard, J. K., Gunter, C.
  2019

- **Trans Effects on Gene Expression Can Drive Omnigenic Inheritance.** *Cell*
  Liu, X., Li, Y. I., Pritchard, J. K.
  2019; 177 (4): 1022

- **Reduced signal for polygenic adaptation of height in UK Biobank** *ELIFE*
  Berg, J. J., Harpak, A., Sinnott-Armstrong, N., Joergensen, A., Mostafavi, H., Field, Y., Boyle, E., Zhang, X., Racimo, F., Pritchard, J. K., Coop, G.
  2019; 8

- **Interpreting polygenic scores, polygenic adaptation, and human phenotypic differences.** *Evolution, medicine, and public health*
  Rosenberg, N. A., Edge, M. D., Pritchard, J. K., Feldman, M. W.
  2019; 2019 (1): 26–34

- **Interpreting polygenic scores, polygenic adaptation, and human phenotypic differences** *EVOLUTION MEDICINE AND PUBLIC HEALTH*
  Rosenberg, N. A., Edge, M. D., Pritchard, J. K., Feldman, M. W.
  2019: 26–34

- **Ancient Rome: A genetic crossroads of Europe and the Mediterranean.** *Science (New York, N.Y.)*
  Antonio, M. L., Gao, Z. n., Moots, H. M., Lucci, M. n., Candilio, F. n., Sawyer, S. n., Oberreiter, V. n., Calderon, D. n., Devitofranceschi, K. n., Aikens, R. C., Aneli, S. n., Bartoli, F. n., Bedini, et al
  2019; 366 (6466): 708–14

- **High-resolution mapping of cancer cell networks using co-functional interactions.** *Molecular systems biology*
  Boyle, E. A., Pritchard, J. K., Greenleaf, W. J.
  2018; 14 (12): e8594

- **Evidence for Weak Selective Constraint on Human Gene Expression.** *Genetics*
  Glassberg, E. C., Gao, Z., Harpak, A., Lan, X., Pritchard, J. K.
  2018

- **Functional Genetic Variants Revealed by Massively Parallel Precise Genome Editing.** *Cell*
  Sharon, E., Chen, S. A., Khosla, N. M., Smith, J. D., Pritchard, J. K., Fraser, H. B.
  2018

- **Post-translational buffering leads to convergent protein expression levels between primates** *GENOME BIOLOGY*
  Wang, S. H., Hsiao, C., Khan, Z., Pritchard, J. K.
  2018; 19: 83
- **Determining the genetic basis of anthracycline-cardiotoxicity by response QTL mapping in induced cardiomyocytes.** *eLife*
  Knowles, D. A., Burrows, C. K., Blischak, J. D., Patterson, K. M., Serie, D. J., Norton, N., Ober, C., Pritchard, J. K., Gilad, Y.
  2018; 7
- **Remodeling the Specificity of an Endosomal CORVET Tether Underlies Formation of Regulated Secretory Vesicles in the Ciliate Tetrahymena thermophila** *CURRENT BIOLOGY*
  Sparvoli, D., Richardson, E., Osakada, H., Lan, X., Iwamoto, M., Bowman, G. R., Kontur, C., Bourland, W. A., Lynn, D. H., Pritchard, J. K., Haraguchi, T., Dacks, J. B., Turkewitz, et al
  2018; 28 (5): 697-+
- **Impact of regulatory variation across human iPSCs and differentiated cells** *GENOME RESEARCH*
  Banovich, N. E., Li, Y. I., Raj, A., Ward, M. C., Greenside, P., Calderon, D., Tung, P., Burnett, J. E., Myrthil, M., Thomas, S. M., Burrows, C. K., Romero, I., Pavlovic, et al
  2018; 28 (1): 122–31
- **Annotation-free quantification of RNA splicing using LeafCutter** *NATURE GENETICS*
  Li, Y. I., Knowles, D. A., Humphrey, J., Barbeira, A. N., Dickinson, S. P., Im, H., Pritchard, J. K.
  2018; 50 (1): 151-+
- **Large-Scale Clonal Analysis Resolves Aging of the Mouse Hematopoietic Stem Cell Compartment.** *Cell stem cell*
  Yamamoto, R. n., Wilkinson, A. C., Ooehara, J. n., Lan, X. n., Lai, C. Y., Nakauchi, Y. n., Pritchard, J. K., Nakauchi, H. n.
  2018; 22 (4): 600–607.e4
- **Frequent nonallelic gene conversion on the human lineage and its effect on the divergence of gene duplicates** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*
  Harpak, A., Lan, X., Gao, Z., Pritchard, J. K.
  2017; 114 (48): 12779–84
- **Inferring Relevant Cell Types for Complex Traits by Using Single-Cell Gene Expression** *AMERICAN JOURNAL OF HUMAN GENETICS*
  Calderon, D., Bhaskar, A., Knowles, D. A., Golan, D., Raj, T., Fu, A. Q., Pritchard, J. K.
  2017; 101 (5): 686–99
- **Quantification of transplant-derived circulating cell-free DNA in absence of a donor genotype** *PLOS COMPUTATIONAL BIOLOGY*
  Sharon, E., Shi, H., Kharbanda, S., Koh, W., Martin, L. R., Khush, K. K., Valantine, H., Pritchard, J. K., De Vlaminck, I.
  2017; 13 (8): e1005629
- **An Expanded View of Complex Traits: From Polygenic to Omnigenic** *CELL*
  Boyle, E. A., Li, Y. I., Pritchard, J. K.
  2017; 169 (7): 1177–86
- **Rapid evolution of the human mutation spectrum** *ELIFE*
  Harris, K., Pritchard, J. K.
  2017; 6
- **Tracing the peopling of the world through genomics.** *Nature*
  Nielsen, R., Akey, J. M., Jakobsson, M., Pritchard, J. K., Tishkoff, S., Willerslev, E.
  2017; 541 (7637): 302-310
- **Batch effects and the effective design of single-cell gene expression studies** *SCIENTIFIC REPORTS*
  Tung, P., Blischak, J. D., Hsiao, C. J., Knowles, D. A., Burnett, J. E., Pritchard, J. K., Gilad, Y.
  2017; 7
- **Mutation Rate Variation is a Primary Determinant of the Distribution of Allele Frequencies in Humans** *PLOS GENETICS*
  Harpak, A., Bhaskar, A., Pritchard, J. K.
  2016; 12 (12)

- **A Bibliometric History of the Journal GENETICS** *GENETICS*
  Telis, N., Lehmann, B. V., Feldman, M. W., Pritchard, J. K.
  2016; 204 (4): 1337-1342
- **Detection of human adaptation during the past 2000 years.** *Science*
  Field, Y., Boyle, E. A., Telis, N., Gao, Z., Gaulton, K. J., Golan, D., Yengo, L., Rocheleau, G., Froguel, P., McCarthy, M. I., Pritchard, J. K.
  2016
- **Lineage-specific and single-cell chromatin accessibility charts human hematopoiesis and leukemia evolution.** *Nature genetics*
  Corces, M. R., Buenrostro, J. D., Wu, B., Greenside, P. G., Chan, S. M., Koenig, J. L., Snyder, M. P., Pritchard, J. K., Kundaje, A., Greenleaf, W. J., Majeti, R., Chang, H. Y.
  2016; 48 (10): 1193-1203
- **Genetic variation in MHC proteins is associated with T cell receptor expression biases.** *Nature genetics*
  Sharon, E., Sibener, L. V., Battle, A., Fraser, H. B., Garcia, K. C., Pritchard, J. K.
  2016; 48 (9): 995-1002
- **Genome-wide association study of behavioral, physiological and gene expression traits in outbred CFW mice.** *Nature genetics*
  Parker, C. C., Gopalakrishnan, S., Carbonetto, P., Gonzales, N. M., Leung, E., Park, Y. J., Aryee, E., Davis, J., Blizard, D. A., Ackert-Bicknell, C. L., Lionikas, A., Pritchard, J. K., Palmer, et al
  2016; 48 (8): 919-926
- **Thousands of novel translated open reading frames in humans inferred by ribosome footprint profiling.** *eLife*
  Raj, A., Wang, S. H., Shim, H., Harpak, A., Li, Y. I., Engelmann, B., Stephens, M., Gilad, Y., Pritchard, J. K.
  2016; 5
- **Coregulation of tandem duplicate genes slows evolution of subfunctionalization in mammals** *SCIENCE*
  Lan, X., Pritchard, J. K.
  2016; 352 (6288): 1009-1013
- **RNA splicing is a primary link between genetic variation and disease** *SCIENCE*
  Li, Y. I., van de Geijn, B., Raj, A., Knowles, D. A., Petti, A. A., Golan, D., Gilad, Y., Pritchard, J. K.
  2016; 352 (6285): 600-604
- **Genetic Variation, Not Cell Type of Origin, Underlies the Majority of Identifiable Regulatory Differences in iPSCs.** *PLoS genetics*
  Burrows, C. K., Banovich, N. E., Pavlovic, B. J., Patterson, K., Gallego Romero, I., Pritchard, J. K., Gilad, Y.
  2016; 12 (1)
- **Abundant contribution of short tandem repeats to gene expression variation in humans.** *Nature genetics*
  Gymrek, M., Willems, T., Guilmatre, A., Zeng, H., Markus, B., Georgiev, S., Daly, M. J., Price, A. L., Pritchard, J. K., Sharp, A. J., Erlich, Y.
  2016; 48 (1): 22-9
- **Genetic Variation, Not Cell Type of Origin, Underlies the Majority of Identifiable Regulatory Differences in iPSCs** *PLOS GENETICS*
  Burrows, C. K., Banovich, N. E., Pavlovic, B. J., Patterson, K., Romero, I. G., Pritchard, J. K., Gilad, Y.
  2016; 12 (1)
- **Abundant contribution of short tandem repeats to gene expression variation in humans** *NATURE GENETICS*
  Gymrek, M., Willems, T., Guilmatre, A., Zeng, H., Markus, B., Georgiev, S., Daly, M. J., Price, A. L., Pritchard, J. K., Sharp, A. J., Erlich, Y.
  2016; 48 (1): 22-?
- **Whole Genome Sequencing Identifies a Novel Factor Required for Secretory Granule Maturation in Tetrahymena thermophila.** *G3 (Bethesda, Md.)*
  Kontur, C., Kumar, S., Lan, X., Pritchard, J. K., Turkewitz, A. P.
  2016; 6 (8): 2505-2516
- **WASP: allele-specific software for robust molecular quantitative trait locus discovery** *NATURE METHODS*
  van de Geijn, B., McVicker, G., Gila, Y., Pritchard, J. K.
  2015; 12 (11): 1061-1063
- **WASP: allele-specific software for robust molecular quantitative trait locus discovery.** *Nature methods*

van de Geijn, B., McVicker, G., Gilad, Y., Pritchard, J. K.
2015; 12 (11): 1061-3

- **Genetic Control of Chromatin States in Humans Involves Local and Distal Chromosomal Interactions** *CELL*
  Grubert, F., Zaugg, J. B., Kasowski, M., Ursu, O., Spacek, D. V., Martin, A. R., Greenside, P., Srivas, R., Phanstiel, D. H., Pekowska, A., Heidari, N., Euskirchen, G., Huber, et al
  2015; 162 (5): 1051-1065
- **Genetic Control of Chromatin States in Humans Involves Local and Distal Chromosomal Interactions.** *Cell*
  Grubert, F., Zaugg, J. B., Kasowski, M., Ursu, O., Spacek, D. V., Martin, A. R., Greenside, P., Srivas, R., Phanstiel, D. H., Pekowska, A., Heidari, N., Euskirchen, G., Huber, et al
  2015; 162 (5): 1051-1065
- **The Genotype-Tissue Expression (GTEx) pilot analysis: Multitissue gene regulation in humans** *SCIENCE*
  Ardlie, K. G., DeLuca, D. S., Segre, A. V., Sullivan, T. J., Young, T. R., Gelfand, E. T., Trowbridge, C. A., Maller, J. B., Tukiainen, T., Lek, M., Ward, L. D., Kheradpour, P., Iriarte, et al
  2015; 348 (6235): 648-660
- **Effect of predicted protein-truncating genetic variants on the human transcriptome** *SCIENCE*
  Rivas, M. A., Pirinen, M., Conrad, D. F., Lek, M., Tsang, E. K., Karczewski, K. J., Maller, J. B., Kukurba, K. R., DeLuca, D. S., Fromer, M., Ferreira, P. G., Smith, K. S., Zhang, et al
  2015; 348 (6235): 666-669
- **Reprogramming LCLs to iPSCs Results in Recovery of Donor-Specific Gene Expression Signature** *PLOS GENETICS*
  Thomas, S. M., Kagan, C., Pavlovic, B. J., Burnett, J., Patterson, K., Pritchard, J. K., Gilad, Y.
  2015; 11 (5)
- **Sharing and Specificity of Co-expression Networks across 35 Human Tissues.** *PLoS computational biology*
  Pierson, E., Koller, D., Battle, A., Mostafavi, S., Ardlie, K. G., Getz, G., Wright, F. A., Kellis, M., Volpi, S., Dermitzakis, E. T.
  2015; 11 (5)
- **Genomic variation. Impact of regulatory variation from RNA to protein.** *Science*
  Battle, A., Khan, Z., Wang, S. H., Mitrano, A., Ford, M. J., Pritchard, J. K., Gilad, Y.
  2015; 347 (6222): 664-667
- **Impact of regulatory variation from RNA to protein** *SCIENCE*
  Battle, A., Khan, Z., Wang, S. H., Mitrano, A., Ford, M. J., Pritchard, J. K., Gilad, Y.
  2015; 347 (6222): 664-667
- **The Genetic and Mechanistic Basis for Variation in Gene Regulation** *PLOS GENETICS*
  Pai, A. A., Pritchard, J. K., Gilad, Y.
  2015; 11 (1)
- **msCentipede: Modeling Heterogeneity across Genomic Sites and Replicates Improves Accuracy in the Inference of Transcription Factor Binding.** *PloS one*
  Raj, A., Shim, H., Gilad, Y., Pritchard, J. K., Stephens, M.
  2015; 10 (9): e0138030
- **msCentipede: Modeling Heterogeneity across Genomic Sites and Replicates Improves Accuracy in the Inference of Transcription Factor Binding.** *PloS one*
  Raj, A., Shim, H., Gilad, Y., Pritchard, J. K., Stephens, M.
  2015; 10 (9)
- **Methylation QTLs Are Associated with Coordinated Changes in Transcription Factor Binding, Histone Modifications, and Gene Expression Levels** *PLOS GENETICS*
  Banovich, N. E., Lan, X., McVicker, G., van de Geijn, B., Degner, J. F., Blischak, J. D., Roux, J., Pritchard, J. K., Gilad, Y.
  2014; 10 (9)
- **Methylation QTLs are associated with coordinated changes in transcription factor binding, histone modifications, and gene expression levels.** *PLoS genetics*
  Banovich, N. E., Lan, X., McVicker, G., van de Geijn, B., Degner, J. F., Blischak, J. D., Roux, J., Pritchard, J. K., Gilad, Y.

2014; 10 (9): e1004663

- **fastSTRUCTURE: Variational Inference of Population Structure in Large SNP Data Sets** *GENETICS*
  Raj, A., Stephens, M., Pritchard, J. K.
  2014; 197 (2): 573-U207
- **fastSTRUCTURE: variational inference of population structure in large SNP data sets.** *Genetics*
  Raj, A., Stephens, M., Pritchard, J. K.
  2014; 197 (2): 573-89
- **The deleterious mutation load is insensitive to recent population history.** *Nature genetics*
  Simons, Y. B., Turchin, M. C., Pritchard, J. K., Sella, G.
  2014; 46 (3): 220-224
- **The functional consequences of variation in transcription factor binding.** *PLoS genetics*
  Cusanovich, D. A., Pavlovic, B., Pritchard, J. K., Gilad, Y.
  2014; 10 (3)
- **The chromatin architectural proteins HMGD1 and H1 bind reciprocally and have opposite effects on chromatin structure and gene regulation** *BMC GENOMICS*
  Nalabothula, N., McVicker, G., Maiorano, J., Martin, R., Pritchard, J. K., Fondufe-Mittendorf, Y. N.
  2014; 15
- **Archaic humans: Four makes a party.** *Nature*
  Birney, E., Pritchard, J. K.
  2014; 505 (7481): 32-4
- **The effect of freeze-thaw cycles on gene expression levels in lymphoblastoid cell lines.** *PloS one*
  Çaliskan, M., Pritchard, J. K., Ober, C., Gilad, Y.
  2014; 9 (9)
- **Epigenetic modifications are associated with inter-species gene expression variation in primates** *GENOME BIOLOGY*
  Zhou, X., Cain, C. E., Myrthil, M., Lewellen, N., Michelini, K., Davenport, E. R., Stephens, M., Pritchard, J. K., Gilad, Y.
  2014; 15 (12)
- **Primate Transcript and Protein Expression Levels Evolve Under Compensatory Selection Pressures** *SCIENCE*
  Khan, Z., Ford, M. J., Cusanovich, D. A., Mitrano, A., Pritchard, J. K., Gilad, Y.
  2013; 342 (6162): 1100-1104
- **Identification of Genetic Variants That Affect Histone Modifications in Human Cells** *SCIENCE*
  McVicker, G., van de Geijn, B., Degner, J. F., Cain, C. E., Banovich, N. E., Raj, A., Lewellen, N., Myrthil, M., Gilad, Y., Pritchard, J. K.
  2013; 342 (6159): 747-749
- **The Genotype-Tissue Expression (GTEx) project** *NATURE GENETICS*
  Lonsdale, J., Thomas, J., Salvatore, M., Phillips, R., Lo, E., Shad, S., Hasz, R., Walters, G., Garcia, F., Young, N., Foster, B., Moser, M., Karasik, et al
  2013; 45 (6): 580-585