

6 References

- 1000 Genomes Project Consortium, Adam Auton, Lisa D. Brooks, Richard M. Durbin, Erik P. Garrison, Hyun Min Kang, Jan O. Korbel, et al. 2015. "A Global Reference for Human Genetic Variation." *Nature* 526 (7571): 68–74.
- Adzhubei, Ivan A., Steffen Schmidt, Leonid Peshkin, Vasily E. Ramensky, Anna Gerasimova, Peer Bork, Alexey S. Kondrashov, and Shamil R. Sunyaev. 2010. "A Method and Server for Predicting Damaging Missense Mutations." *Nature Methods* 7 (4): 248–49.
- Anders, Simon, Paul Theodor Pyl, and Wolfgang Huber. 2015. "HTSeq--a Python Framework to Work with High-Throughput Sequencing Data." *Bioinformatics* 31 (2): 166–69.
- Andersson, Robin, Claudia Gebhard, Irene Miguel-Escalada, Ilka Hoof, Jette Bornholdt, Mette Boyd, Yun Chen, et al. 2014. "An Atlas of Active Enhancers across Human Cell Types and Tissues." *Nature* 507 (7493): 455–61.
- Ashley, A. K., A. I. Hinds, W. H. Hanneman, R. B. Tjalkens, and M. E. Legare. 2016. "DJ-1 Mutation Decreases Astroglial Release of Inflammatory Mediators." *Neurotoxicology* 52 (January): 198–203.
- Barbeira, Alvaro, Scott P. Dickinson, Jason M. Torres, Rodrigo Bonazzola, Jiamao Zheng, Eric S. Torstenson, Heather E. Wheeler, et al. 2017. "Integrating Tissue Specific Mechanisms into GWAS Summary Results." *bioRxiv*. doi:10.1101/045260.
- Bardy, C., M. van den Hurk, B. Kakaradov, J. A. Erwin, B. N. Jaeger, R. V. Hernandez, T. Eames, et al. 2016. "Predicting the Functional States of Human iPSC-Derived Neurons with Single-Cell RNA-Seq and Electrophysiology." *Molecular Psychiatry* 21 (11): 1573–88.
- Bar-Nur, Ori, Holger A. Russ, Shimon Efrat, and Nissim Benvenisty. 2011. "Epigenetic Memory and Preferential Lineage-Specific Differentiation in Induced Pluripotent Stem Cells Derived from Human Pancreatic Islet Beta Cells." *Cell Stem Cell* 9 (1): 17–23.
- Baron, Maayan, Adrian Veres, Samuel L. Wolock, Aubrey L. Faust, Renaud Gaujoux, Amedeo Vetere, Jennifer Hyoje Ryu, et al. 2016. "A Single-Cell Transcriptomic Map of the Human and Mouse Pancreas Reveals Inter- and Intra-Cell Population Structure." *Cell Systems* 3 (4): 346–60.e4.
- Barski, Artem, Suresh Cuddapah, Kairong Cui, Tae-Young Roh, Dustin E. Schones, Zhibin Wang, Gang Wei, Iouri Chepelev, and Keji Zhao. 2007. "High-Resolution Profiling of Histone Methylation in the Human Genome." *Cell* 129 (4): 823–37.
- Battle, Alexis, Zia Khan, Sidney H. Wang, Amy Mitrano, Michael J. Ford, Jonathan K. Pritchard, and Yoav Gilad. 2015. "Genomic Variation. Impact of Regulatory Variation from RNA to Protein." *Science* 347 (6222): 664–67.
- Battle, Alexis, Sara Mostafavi, Xiaowei Zhu, James B. Potash, Myrna M. Weissman, Courtney McCormick, Christian D. Haudenschild, et al. 2014. "Characterizing the Genetic Basis of Transcriptome Diversity through RNA-Sequencing of 922 Individuals." *Genome Research* 24 (1): 14–24.
- Benjamini, Yuval, and Terence P. Speed. 2012. "Summarizing and Correcting the GC Content Bias in High-Throughput Sequencing." *Nucleic Acids Research* 40 (10): e72.
- Benner, Christian, Chris C. A. Spencer, Aki S. Havulinna, Veikko Salomaa, Samuli Ripatti, and Matti Pirinen. 2016. "FINEMAP: Efficient Variable Selection Using Summary Data from Genome-Wide Association Studies." *Bioinformatics* 32 (10): 1493–1501.
- Bielekova, B., M. Catalfamo, S. Reichert-Scrivner, A. Packer, M. Cerna, T. A. Waldmann, H. McFarland, P. A. Henkart, and R. Martin. 2006. "Regulatory CD56bright Natural Killer Cells Mediate Immunomodulatory Effects of IL-2R -Targeted Therapy (daclizumab) in Multiple Sclerosis." *Proceedings of the National Academy of Sciences* 103 (15): 5941–46.

- Bock, Christoph, Evangelos Kiskinis, Griet Verstappen, Hongcang Gu, Gabriella Boultling, Zachary D. Smith, Michael Ziller, et al. 2011. "Reference Maps of Human ES and iPS Cell Variation Enable High-Throughput Characterization of Pluripotent Cell Lines." *Cell* 144 (3): 439–52.
- Bojesen, Stig E., Karen A. Pooley, Sharon E. Johnatty, Jonathan Beesley, Kyriaki Michailidou, Jonathan P. Tyrer, Stacey L. Edwards, et al. 2013. "Multiple Independent Variants at the TERT Locus Are Associated with Telomere Length and Risks of Breast and Ovarian Cancer." *Nature Genetics* 45 (4): 371–84, 384e1–2.
- Boyle, Alan P., Eurie L. Hong, Manoj Hariharan, Yong Cheng, Marc A. Schaub, Maya Kasowski, Konrad J. Karczewski, et al. 2012. "Annotation of Functional Variation in Personal Genomes Using RegulomeDB." *Genome Research* 22 (9): 1790–97.
- Buenrostro, Jason D., Paul G. Giresi, Lisa C. Zaba, Howard Y. Chang, and William J. Greenleaf. 2013. "Transposition of Native Chromatin for Fast and Sensitive Epigenomic Profiling of Open Chromatin, DNA-Binding Proteins and Nucleosome Position." *Nature Methods* 10 (12): 1213–18.
- Bunt, Martijn van de, Adrian Cortes, IGAS Consortium, Matthew A. Brown, Andrew P. Morris, and Mark I. McCarthy. 2015. "Evaluating the Performance of Fine-Mapping Strategies at Common Variant GWAS Loci." *PLoS Genetics* 11 (9): e1005535.
- Burrows, Courtney K., Nicholas E. Banovich, Bryan J. Pavlovic, Kristen Patterson, Irene Gallego Romero, Jonathan K. Pritchard, and Yoav Gilad. 2016. "Genetic Variation, Not Cell Type of Origin, Underlies the Majority of Identifiable Regulatory Differences in iPSCs." *PLoS Genetics* 12 (1): e1005793.
- Cacchiarelli, Davide, Xiaojie Qiu, Sanjay Srivatsan, Michael Ziller, Eliah Overbey, Jonna Grimsby, Prapti Pokharel, et al. 2017. "Aligning Single-Cell Developmental and Reprogramming Trajectories Identifies Molecular Determinants of Reprogramming Outcome." *bioRxiv*. doi:10.1101/122531.
- Cao, Lishuang, Aoibhinn McDonnell, Anja Nitzsche, Aristos Alexandrou, Pierre-Philippe Saintot, Alexandre J. C. Loucif, Adam R. Brown, et al. 2016. "Pharmacological Reversal of a Pain Phenotype in iPSC-Derived Sensory Neurons and Patients with Inherited Erythromelalgia." *Science Translational Medicine* 8 (335): 335ra56.
- Carbone, Fortunata, Veronica De Rosa, Pietro B. Carrieri, Silvana Montella, Dario Bruzzese, Antonio Porcellini, Claudio Procaccini, Antonio La Cava, and Giuseppe Matarese. 2014. "Regulatory T Cell Proliferative Potential Is Impaired in Human Autoimmune Disease." *Nature Medicine* 20 (1): 69–74.
- Castel, Stephane E., Ami Levy-Moonshine, Pejman Mohammadi, Eric Banks, and Tuuli Lappalainen. 2015. "Tools and Best Practices for Data Processing in Allelic Expression Analysis." *Genome Biology* 16 (September): 195.
- Chambers, Stuart M., Yuchen Qi, Yvonne Mica, Gabsang Lee, Xin-Jun Zhang, Lei Niu, James Bilsland, et al. 2012. "Combined Small-Molecule Inhibition Accelerates Developmental Timing and Converts Human Pluripotent Stem Cells into Nociceptors." *Nature Biotechnology* 30 (7): 715–20.
- Chen, Wenan, Beth R. Larrabee, Inna G. Ovsyannikova, Richard B. Kennedy, Iana H. Haralambieva, Gregory A. Poland, and Daniel J. Schaid. 2015. "Fine Mapping Causal Variants with an Approximate Bayesian Method Using Marginal Test Statistics." *Genetics* 200 (3): 719–36.
- Chong, Jessica X., Kati J. Buckingham, Shalini N. Jhangiani, Corinne Boehm, Nara Sobreira, Joshua D. Smith, Tanya M. Harrell, et al. 2015. "The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities." *American Journal of Human Genetics* 97 (2): 199–215.

- Chronis, Constantinos, Petko Fiziev, Bernadett Papp, Stefan Butz, Giancarlo Bonora, Shan Sabri, Jason Ernst, and Kathrin Plath. 2017. "Cooperative Binding of Transcription Factors Orchestrates Reprogramming." *Cell* 168 (3): 442–59.e20.
- Chun, Sung, Alexandra Casparino, Nikolaos A. Patsopoulos, Damien C. Croteau-Chonka, Benjamin A. Raby, Philip L. De Jager, Shamil R. Sunyaev, and Chris Cotsapas. 2017. "Limited Statistical Evidence for Shared Genetic Effects of eQTLs and Autoimmune-Disease-Associated Loci in Three Major Immune-Cell Types." *Nature Genetics* 49 (4): 600–605.
- Church, Chris, Lee Moir, Fiona McMurray, Christophe Girard, Gareth T. Banks, Lydia Teboul, Sara Wells, et al. 2010. "Overexpression of Fto Leads to Increased Food Intake and Results in Obesity." *Nature Genetics* 42 (12): 1086–92.
- Cingolani, Pablo, Adrian Platts, Le Lily Wang, Melissa Coon, Tung Nguyen, Luan Wang, Susan J. Land, Xiangyi Lu, and Douglas M. Ruden. 2012. "A Program for Annotating and Predicting the Effects of Single Nucleotide Polymorphisms, SnpEff: SNPs in the Genome of Drosophila Melanogaster Strain w1118; Iso-2; Iso-3." *Fly* 6 (2): 80–92.
- Claussnitzer, Melina, Simon N. Dankel, Kyoung-Han Kim, Gerald Quon, Wouter Meuleman, Christine Haugen, Viktoria Glunk, et al. 2015. "FTO Obesity Variant Circuitry and Adipocyte Browning in Humans." *The New England Journal of Medicine* 373 (10): 895–907.
- Creyghton, Menno P., Albert W. Cheng, G. Grant Welstead, Tristan Kooistra, Bryce W. Carey, Eveline J. Steine, Jacob Hanna, et al. 2010. "Histone H3K27ac Separates Active from Poised Enhancers and Predicts Developmental State." *Proceedings of the National Academy of Sciences of the United States of America* 107 (50): 21931–36.
- Danecek, Petr, Adam Auton, Goncalo Abecasis, Cornelis A. Albers, Eric Banks, Mark A. DePristo, Robert E. Handsaker, et al. 2011. "The Variant Call Format and VCFtools." *Bioinformatics* 27 (15): 2156–58.
- D'Antonio, Matteo, Grace Woodruff, Jason L. Nathanson, Agnieszka D'Antonio-Chronowska, Angelo Arias, Hiroko Matsui, Roy Williams, et al. 2017. "High-Throughput and Cost-Effective Characterization of Induced Pluripotent Stem Cells." *Stem Cell Reports*, April. doi:10.1016/j.stemcr.2017.03.011.
- Davis, Joe R., Laure Fresard, David A. Knowles, Mauro Pala, Carlos D. Bustamante, Alexis Battle, and Stephen B. Montgomery. 2016. "An Efficient Multiple-Testing Adjustment for eQTL Studies That Accounts for Linkage Disequilibrium between Variants." *American Journal of Human Genetics* 98 (1): 216–24.
- Davydov, Eugene V., David L. Goode, Marina Sirota, Gregory M. Cooper, Arend Sidow, and Serafim Batzoglou. 2010. "Identifying a High Fraction of the Human Genome to Be under Selective Constraint Using GERP." *PLoS Computational Biology* 6 (12): e1001025.
- Degner, Jacob F., Athma A. Pai, Roger Pique-Regi, Jean-Baptiste Veyrieras, Daniel J. Gaffney, Joseph K. Pickrell, Sherryl De Leon, et al. 2012. "DNase I Sensitivity QTLs Are a Major Determinant of Human Expression Variation." *Nature* 482 (7385): 390–94.
- DeLuca, David S., Joshua Z. Levin, Andrey Sivachenko, Timothy Fennell, Marc-Danie Nazaire, Chris Williams, Michael Reich, Wendy Winckler, and Gad Getz. 2012. "RNA-SeQC: RNA-Seq Metrics for Quality Control and Process Optimization." *Bioinformatics* 28 (11): 1530–32.
- Dianat, Noushin, Clara Steichen, Ludovic Vallier, Anne Weber, and Anne Dubart-Kupperschmitt. 2013. "Human Pluripotent Stem Cells for Modelling Human Liver Diseases and Cell Therapy." *Current Gene Therapy* 13 (2): 120–32.
- Dimas, A. S., S. Deutsch, B. E. Stranger, S. B. Montgomery, C. Borel, H. Attar-Cohen, C. Ingle, et al. 2009. "Common Regulatory Variation Impacts Gene Expression in a Cell Type-Dependent Manner." *Science* 325 (5945): 1246–50.

- Ding, Jun, Johann E. Gudjonsson, Liming Liang, Philip E. Stuart, Yun Li, Wei Chen, Michael Weichenthal, et al. 2010. "Gene Expression in Skin and Lymphoblastoid Cells: Refined Statistical Method Reveals Extensive Overlap in Cis-eQTL Signals." *American Journal of Human Genetics* 87 (6): 779–89.
- Dixon, Jesse R., Siddarth Selvaraj, Feng Yue, Audrey Kim, Yan Li, Yin Shen, Ming Hu, Jun S. Liu, and Bing Ren. 2012. "Topological Domains in Mammalian Genomes Identified by Analysis of Chromatin Interactions." *Nature* 485 (7398): 376–80.
- Doniger, Scott W., and Justin C. Fay. 2007. "Frequent Gain and Loss of Functional Transcription Factor Binding Sites." *PLoS Computational Biology* 3 (5): e99.
- Dunham, Ian, Eugene Kulesha, Valentina Iotchkova, Sandro Morganella, and Ewan Birney. 2015. "FORGE: A Tool to Discover Cell Specific Enrichments of GWAS Associated SNPs in Regulatory Regions." *F1000Research*. doi:10.12688/f1000research.6032.1.
- ENCODE Project Consortium. 2012. "An Integrated Encyclopedia of DNA Elements in the Human Genome." *Nature* 489 (7414): 57–74.
- Ernst, Jason, and Manolis Kellis. 2012. "ChromHMM: Automating Chromatin-State Discovery and Characterization." *Nature Methods* 9 (3): 215–16.
- Ernst, Jason, and Manolis Kellis. 2015. "Large-Scale Imputation of Epigenomic Datasets for Systematic Annotation of Diverse Human Tissues." *Nature Biotechnology* 33 (4): 364–76.
- Evans, David M., Chris C. A. Spencer, Jennifer J. Pointon, Zhan Su, David Harvey, Grazyna Kochan, Udo Oppermann, et al. 2011. "Interaction between ERAP1 and HLA-B27 in Ankylosing Spondylitis Implicates Peptide Handling in the Mechanism for HLA-B27 in Disease Susceptibility." *Nature Genetics* 43 (8): 761–67.
- Fairfax, Benjamin P., Peter Humburg, Seiko Makino, Vivek Naranbhai, Daniel Wong, Evelyn Lau, Luke Jostins, et al. 2014. "Innate Immune Activity Conditions the Effect of Regulatory Variants upon Monocyte Gene Expression." *Science* 343 (6175): 1246949.
- FANTOM Consortium and the RIKEN PMI and CLST (DGT), Alistair R. R. Forrest, Hideya Kawaji, Michael Rehli, J. Kenneth Baillie, Michiel J. L. de Hoon, Vanja Haberle, et al. 2014. "A Promoter-Level Mammalian Expression Atlas." *Nature* 507 (7493): 462–70.
- Farh, Kyle Kai-How, Alexander Marson, Jiang Zhu, Markus Kleinewietfeld, William J. Housley, Samantha Beik, Noam Shoresh, et al. 2015. "Genetic and Epigenetic Fine Mapping of Causal Autoimmune Disease Variants." *Nature* 518 (7539): 337–43.
- Ferraro, A., A. M. D'Alise, T. Raj, N. Asinovski, R. Phillips, A. Ergun, J. M. Replogle, et al. 2014. "Interindividual Variation in Human T Regulatory Cells." *Proceedings of the National Academy of Sciences* 111 (12): E1111–20.
- Finucane, Hilary K., Brendan Bulik-Sullivan, Alexander Gusev, Gosia Trynka, Yakir Reshef, Po-Ru Loh, Verner Anttila, et al. 2015. "Partitioning Heritability by Functional Annotation Using Genome-Wide Association Summary Statistics." *Nature Genetics* 47 (11): 1228–35.
- Fischer, Julia, Linda Koch, Christian Emmerling, Jeanette Vierkotten, Thomas Peters, Jens C. Brüning, and Ulrich Rüther. 2009. "Inactivation of the Fto Gene Protects from Obesity." *Nature* 458 (7240): 894–98.
- Fuchsberger, Christian, Jason Flannick, Tanya M. Teslovich, Anubha Mahajan, Vineeta Agarwala, Kyle J. Gaulton, Clement Ma, et al. 2016. "The Genetic Architecture of Type 2 Diabetes." *Nature* 536 (7614): 41–47.
- Gaffney, Daniel J., Jean-Baptiste Veyrieras, Jacob F. Degner, Roger Pique-Regi, Athma A. Pai, Gregory E. Crawford, Matthew Stephens, Yoav Gilad, and Jonathan K. Pritchard. 2012. "Dissecting the Regulatory Architecture of Gene Expression QTLs." *Genome Biology* 13 (1): R7.
- Gagliano, Sarah A., Michael R. Barnes, Michael E. Weale, and Jo Knight. 2014. "A Bayesian Method to Incorporate Hundreds of Functional Characteristics with Association Evidence to Improve Variant Prioritization." *PLoS One* 9 (5): e98122.

- Gamazon, Eric R., Heather E. Wheeler, Kaanan P. Shah, Sahar V. Mozaffari, Keston Aquino-Michaels, Robert J. Carroll, Anne E. Eyler, et al. 2015. "A Gene-Based Association Method for Mapping Traits Using Reference Transcriptome Data." *Nature Genetics* 47 (9): 1091–98.
- Gates, Leah A., Jiejun Shi, Aarti D. Rohira, Qin Feng, Bokai Zhu, Mark T. Bedford, Cari A. Sagum, et al. 2017. "Acetylation on Histone H3 Lysine 9 Mediates a Switch from Transcription Initiation to Elongation." *The Journal of Biological Chemistry* 292 (35): 14456–72.
- Ghanbari, Mohsen, M. Arfan Ikram, Hans W. J. de Looper, Albert Hofman, Stefan J. Erkeland, Oscar H. Franco, and Abbas Dehghan. 2016. "Genome-Wide Identification of microRNA-Related Variants Associated with Risk of Alzheimer's Disease." *Scientific Reports* 6 (1). doi:10.1038/srep28387.
- Giambartolomei, Claudia, Damjan Vukcevic, Eric E. Schadt, Lude Franke, Aroon D. Hingorani, Chris Wallace, and Vincent Plagnol. 2014. "Bayesian Test for Colocalisation between Pairs of Genetic Association Studies Using Summary Statistics." *PLoS Genetics* 10 (5): e1004383.
- Global Lipids Genetics Consortium, Cristen J. Willer, Ellen M. Schmidt, Sebanti Sengupta, Gina M. Peloso, Stefan Gustafsson, Stavroula Kanoni, et al. 2013. "Discovery and Refinement of Loci Associated with Lipid Levels." *Nature Genetics* 45 (11): 1274–83.
- Glubb, Dylan M., Mel J. Maranian, Kyriaki Michailidou, Karen A. Pooley, Kerstin B. Meyer, Siddhartha Kar, Saskia Carlebur, et al. 2015. "Fine-Scale Mapping of the 5q11.2 Breast Cancer Locus Reveals at Least Three Independent Risk Variants Regulating MAP3K1." *American Journal of Human Genetics* 96 (1): 5–20.
- González-Porta, Mar, Adam Frankish, Johan Rung, Jennifer Harrow, and Alvis Brazma. 2013. "Transcriptome Analysis of Human Tissues and Cell Lines Reveals One Dominant Transcript per Gene." *Genome Biology* 14 (7): R70.
- Gregory, Adam P., Calliope A. Dendrou, Kathrine E. Atfield, Aiden Haghikia, Dionysia K. Xifara, Falk Butter, Gereon Poschmann, et al. 2012. "TNF Receptor 1 Genetic Risk Mirrors Outcome of Anti-TNF Therapy in Multiple Sclerosis." *Nature* 488 (7412): 508–11.
- Grzybek, Maciej, Aleksandra Golonko, Marta Walczak, and Paweł Lisowski. 2017. "Epigenetics of Cell Fate Reprogramming and Its Implications for Neurological Disorders Modelling." *Neurobiology of Disease* 99 (March): 84–120.
- GTEx Consortium. 2013. "The Genotype-Tissue Expression (GTEx) Project." *Nature Genetics* 45 (6): 580–85.
- GTEx Consortium. 2015. "Human Genomics. The Genotype-Tissue Expression (GTEx) Pilot Analysis: Multitissue Gene Regulation in Humans." *Science* 348 (6235): 648–60.
- GTEx Consortium et al. 2017. "Genetic Effects on Gene Expression across Human Tissues." *Nature* 550 (7675): 204–13.
- Guenther, Catherine A., Bosiljka Tasic, Liqun Luo, Mary A. Bedell, and David M. Kingsley. 2014. "A Molecular Basis for Classic Blond Hair Color in Europeans." *Nature Genetics* 46 (7): 748–52.
- Gulko, Brad, Melissa J. Hubisz, Ilan Gronau, and Adam Siepel. 2015. "A Method for Calculating Probabilities of Fitness Consequences for Point Mutations across the Human Genome." *Nature Genetics* 47 (3): 276–83.
- Gupta, Rajat M., Joseph Hadaya, Aditi Trehan, Seyedeh M. Zekavat, Carolina Roselli, Derek Klarin, Connor A. Emdin, et al. 2017. "A Genetic Variant Associated with Five Vascular Diseases Is a Distal Regulator of Endothelin-1 Gene Expression." *Cell* 170 (3): 522–33.e15.
- Gusev, Alexander, Arthur Ko, Huwenbo Shi, Gaurav Bhatia, Wonil Chung, Brenda W. J. H.

- Penninx, Rick Jansen, et al. 2016. "Integrative Approaches for Large-Scale Transcriptome-Wide Association Studies." *Nature Genetics* 48 (3): 245–52.
- Hait, Tom Aharon, David Amar, Ron Shamir, and Ran Elkon. 2017. "An Extensive Enhancer-Promoter Map Generated by Genome-Scale Analysis of Enhancer and Gene Activity Patterns." *bioRxiv*. doi:10.1101/190231.
- Handel, Adam E., Satyan Chintawar, Tatjana Lalic, Emma Whiteley, Jane Vowles, Alice Giustacchini, Karen Argoud, et al. 2016. "Assessing Similarity to Primary Tissue and Cortical Layer Identity in Induced Pluripotent Stem Cell-Derived Cortical Neurons through Single-Cell Transcriptomics." *Human Molecular Genetics* 25 (5): 989–1000.
- Hansen, Kasper D., Rafael A. Irizarry, and Zhipin Wu. 2012. "Removing Technical Variability in RNA-Seq Data Using Conditional Quantile Normalization." *Biostatistics* 13 (2): 204–16.
- Harrow, Jennifer, Adam Frankish, Jose M. Gonzalez, Electra Tapanari, Mark Diekhans, Felix Kokocinski, Bronwen L. Aken, et al. 2012. "GENCODE: The Reference Human Genome Annotation for The ENCODE Project." *Genome Research* 22 (9): 1760–74.
- Heinz, Sven, Christopher Benner, Nathanael Spann, Eric Bertolino, Yin C. Lin, Peter Laslo, Jason X. Cheng, Cornelis Murre, Harinder Singh, and Christopher K. Glass. 2010. "Simple Combinations of Lineage-Determining Transcription Factors Prime Cis-Regulatory Elements Required for Macrophage and B Cell Identities." *Molecular Cell* 38 (4): 576–89.
- Hindorff, Lucia A., Praveen Sethupathy, Heather A. Junkins, Erin M. Ramos, Jayashri P. Mehta, Francis S. Collins, and Teri A. Manolio. 2009. "Potential Etiologic and Functional Implications of Genome-Wide Association Loci for Human Diseases and Traits." *Proceedings of the National Academy of Sciences of the United States of America* 106 (23): 9362–67.
- Hoffman, Michael M., Orion J. Buske, Jie Wang, Zhiping Weng, Jeff A. Bilmes, and William Stafford Noble. 2012. "Unsupervised Pattern Discovery in Human Chromatin Structure through Genomic Segmentation." *Nature Methods* 9 (5): 473–76.
- Howie, Bryan, Christian Fuchsberger, Matthew Stephens, Jonathan Marchini, and Gonçalo R. Abecasis. 2012. "Fast and Accurate Genotype Imputation in Genome-Wide Association Studies through Pre-Phasing." *Nature Genetics* 44 (8): 955–59.
- Huang, Hailiang, Ming Fang, Luke Jostins, Maša Umićević Mirkov, Gabrielle Boucher, Carl A. Anderson, Vibeke Andersen, et al. 2017. "Fine-Mapping Inflammatory Bowel Disease Loci to Single-Variant Resolution." *Nature* 547 (7662): 173–78.
- Huang, Yi-Fei, Brad Gulko, and Adam Siepel. 2017. "Fast, Scalable Prediction of Deleterious Noncoding Variants from Functional and Population Genomic Data." *Nature Genetics* 49 (4): 618–24.
- Hu, Bao-Yang, Jason P. Weick, Junying Yu, Li-Xiang Ma, Xiao-Qing Zhang, James A. Thomson, and Su-Chun Zhang. 2010. "Neural Differentiation of Human Induced Pluripotent Stem Cells Follows Developmental Principles but with Variable Potency." *Proceedings of the National Academy of Sciences of the United States of America* 107 (9): 4335–40.
- Hunt, S. P., A. Pini, and G. Evan. 1987. "Induction of c-Fos-like Protein in Spinal Cord Neurons Following Sensory Stimulation." *Nature* 328 (6131): 632–34.
- Inoue, Fumitaka, Martin Kircher, Beth Martin, Gregory M. Cooper, Daniela M. Witten, Michael T. McManus, Nadav Ahituv, and Jay Shendure. 2016. "A Systematic Comparison Reveals Substantial Differences in Chromosomal versus Episomal Encoding of Enhancer Activity." *Genome Research*. doi:10.1101/gr.212092.116.
- Ioannidis, Nilah M., Joe R. Davis, Marianne K. DeGorter, Nicholas B. Larson, Shannon K. McDonnell, Amy J. French, Alexis J. Battle, et al. 2017. "FIRE: Functional Inference of

- Genetic Variants That Regulate Gene Expression." *Bioinformatics* , August. doi:10.1093/bioinformatics/btx534.
- Iotchkova, Valentina, Graham R. S. Ritchie, Matthias Geihs, Sandro Morganella, Josine L. Min, Klaudia Walter, Nicholas J. Timpson, et al. 2016. "GARFIELD - GWAS Analysis of Regulatory or Functional Information Enrichment with LD Correction." *bioRxiv*. doi:10.1101/085738.
- Itzhaki, Ilanit, Leonid Maizels, Irit Huber, Limor Zwi-Dantsis, Oren Caspi, Aaron Winterstern, Oren Feldman, et al. 2011. "Modelling the Long QT Syndrome with Induced Pluripotent Stem Cells." *Nature* 471 (7337): 225–29.
- Iwafuchi-Doi, Makiko, Greg Donahue, Akshay Kakumanu, Jason A. Watts, Shaun Mahony, B. Franklin Pugh, Dolim Lee, Klaus H. Kaestner, and Kenneth S. Zaret. 2016. "The Pioneer Transcription Factor FoxA Maintains an Accessible Nucleosome Configuration at Enhancers for Tissue-Specific Gene Activation." *Molecular Cell* 62 (1): 79–91.
- Iyer, Matthew K., Yashar S. Niknafs, Rohit Malik, Udit Singhal, Anirban Sahu, Yasuyuki Hosono, Terrence R. Barrette, et al. 2015. "The Landscape of Long Noncoding RNAs in the Human Transcriptome." *Nature Genetics* 47 (3): 199–208.
- Javierre, Biola M., Oliver S. Burren, Steven P. Wilder, Roman Kreuzhuber, Steven M. Hill, Sven Sewitz, Jonathan Cairns, et al. 2016. "Lineage-Specific Genome Architecture Links Enhancers and Non-Coding Disease Variants to Target Gene Promoters." *Cell* 167 (5): 1369–84.e19.
- Jordan, J. Dedrick, John Cijiang He, Narat J. Eungdamrong, Ivone Gomes, Wasif Ali, Tracy Nguyen, Trever G. Bivona, Mark R. Philips, Lakshmi A. Devi, and Ravi Iyengar. 2005. "Cannabinoid Receptor-Induced Neurite Outgrowth Is Mediated by Rap1 Activation through G(alpha)o/i-Triggered Proteasomal Degradation of Rap1GAPII." *The Journal of Biological Chemistry* 280 (12): 11413–21.
- Jun, Goo, Matthew Flickinger, Kurt N. Hetrick, Jane M. Romm, Kimberly F. Doheny, Gonçalo R. Abecasis, Michael Boehnke, and Hyun Min Kang. 2012. "Detecting and Estimating Contamination of Human DNA Samples in Sequencing and Array-Based Genotype Data." *American Journal of Human Genetics* 91 (5): 839–48.
- Jung, Seung Hyo, Kyung Jong Won, Kang Pa Lee, Dong Hyun Lee, Suyeol Yu, Dong-Youb Lee, Eun-Hye Seo, et al. 2014. "DJ-1 Protein Regulates CD3+ T Cell Migration via Overexpression of CXCR4 Receptor." *Atherosclerosis* 235 (2): 503–9.
- Kathiresan, Sekar, Olle Melander, Candace Guiducci, Aarti Surti, Noël P. Burtt, Mark J. Rieder, Gregory M. Cooper, et al. 2008. "Six New Loci Associated with Blood Low-Density Lipoprotein Cholesterol, High-Density Lipoprotein Cholesterol or Triglycerides in Humans." *Nature Genetics* 40 (2): 189–97.
- Kelley, David R., Jasper Snoek, and John L. Rinn. 2016. "Basset: Learning the Regulatory Code of the Accessible Genome with Deep Convolutional Neural Networks." *Genome Research* 26 (7): 990–99.
- Kichaev, Gleb, Megan Roytman, Ruth Johnson, Eleazar Eskin, Sara Lindström, Peter Kraft, and Bogdan Pasaniuc. 2017. "Improved Methods for Multi-Trait Fine Mapping of Pleiotropic Risk Loci." *Bioinformatics* 33 (2): 248–55.
- Kichaev, Gleb, Wen-Yun Yang, Sara Lindstrom, Farhad Hormozdiari, Eleazar Eskin, Alkes L. Price, Peter Kraft, and Bogdan Pasaniuc. 2014. "Integrating Functional Data to Prioritize Causal Variants in Statistical Fine-Mapping Studies." *PLoS Genetics* 10 (10): e1004722.
- Kilpinen, Helena, Angela Goncalves, Andreas Leha, Vackar Afzal, Kaur Alasoo, Sofie Ashford, Sendu Bala, et al. 2017. "Common Genetic Variation Drives Molecular Heterogeneity in Human iPSCs." *Nature* 546 (7658): 370–75.
- Kim, K., A. Doi, B. Wen, K. Ng, R. Zhao, P. Cahan, J. Kim, et al. 2010. "Epigenetic Memory

- in Induced Pluripotent Stem Cells." *Nature* 467 (7313): 285–90.
- Kindt, Alida S. D., Pau Navarro, Colin A. M. Semple, and Chris S. Haley. 2013. "The Genomic Signature of Trait-Associated Variants." *BMC Genomics* 14 (February): 108.
- Kircher, Martin, Daniela M. Witten, Preti Jain, Brian J. O'Roak, Gregory M. Cooper, and Jay Shendure. 2014. "A General Framework for Estimating the Relative Pathogenicity of Human Genetic Variants." *Nature Genetics* 46 (3): 310–15.
- Kiselev, Vladimir Yu, Kristina Kirschner, Michael T. Schaub, Tallulah Andrews, Andrew Yiu, Tamir Chandra, Kedar N. Natarajan, et al. 2017. "SC3: Consensus Clustering of Single-Cell RNA-Seq Data." *Nature Methods* 14 (5): 483–86.
- Kléber, Maurice, Hye-Youn Lee, Heiko Wurdak, Johanna Buchstaller, Martin M. Riccomagno, Lars M. Ittner, Ueli Suter, Douglas J. Epstein, and Lukas Sommer. 2005. "Neural Crest Stem Cell Maintenance by Combinatorial Wnt and BMP Signaling." *The Journal of Cell Biology* 169 (2): 309–20.
- Kohno, Tatsuro, Kimberly A. Moore, Hiroshi Baba, and Clifford J. Woolf. 2003. "Peripheral Nerve Injury Alters Excitatory Synaptic Transmission in Lamina II of the Rat Dorsal Horn." *The Journal of Physiology* 548 (Pt 1): 131–38.
- Kolde, Raivo, Sven Laur, Priit Adler, and Jaak Vilo. 2012. "Robust Rank Aggregation for Gene List Integration and Meta-Analysis." *Bioinformatics* 28 (4): 573–80.
- Kumar, Prateek, Steven Henikoff, and Pauline C. Ng. 2009. "Predicting the Effects of Coding Non-Synonymous Variants on Protein Function Using the SIFT Algorithm." *Nature Protocols* 4 (7): 1073–81.
- Kumasaka, Natsuhiko, Andrew J. Knights, and Daniel J. Gaffney. 2016. "Fine-Mapping Cellular QTLs with RASQUAL and ATAC-Seq." *Nature Genetics* 48 (2): 206–13.
- Lahens, Nicholas F., Ibrahim Halil Kavakli, Ray Zhang, Katharina Hayer, Michael B. Black, Hannah Dueck, Angel Pizarro, et al. 2014. "IVT-Seq Reveals Extreme Bias in RNA Sequencing." *Genome Biology* 15 (6): R86.
- Lange, Katrina M. de, Loukas Moutsianas, James C. Lee, Christopher A. Lamb, Yang Luo, Nicholas A. Kennedy, Luke Jostins, et al. 2017. "Genome-Wide Association Study Implicates Immune Activation of Multiple Integrin Genes in Inflammatory Bowel Disease." *Nature Genetics* 49 (2): 256–61.
- Lappalainen, Tuuli, Michael Sammeth, Marc R. Friedländer, Peter A. C. 't Hoen, Jean Monlong, Manuel A. Rivas, Mar González-Porta, et al. 2013. "Transcriptome and Genome Sequencing Uncovers Functional Variation in Humans." *Nature* 501 (7468): 506–11.
- Lee, Dongwon, David U. Gorkin, Maggie Baker, Benjamin J. Strober, Alessandro L. Asoni, Andrew S. McCallion, and Michael A. Beer. 2015. "A Method to Predict the Impact of Regulatory Variants from DNA Sequence." *Nature Genetics* 47 (8): 955–61.
- Lee, Gabsang, Lee Gabsang, Eirini P. Papapetrou, Kim Hyesoo, Stuart M. Chambers, Mark J. Tomishima, Christopher A. Fasano, et al. 2009. "Modelling Pathogenesis and Treatment of Familial Dysautonomia Using Patient-Specific iPSCs." *Nature* 461 (7262): 402–6.
- Lee, Mark N., Chun Ye, Alexandra-Chloé Villani, Towfique Raj, Weibo Li, Thomas M. Eisenhaure, Selina H. Imboywa, et al. 2014. "Common Genetic Variants Modulate Pathogen-Sensing Responses in Human Dendritic Cells." *Science* 343 (6175): 1246980.
- Lee, Seunggeung, Gonçalo R. Abecasis, Michael Boehnke, and Xihong Lin. 2014. "Rare-Variant Association Analysis: Study Designs and Statistical Tests." *American Journal of Human Genetics* 95 (1): 5–23.
- Lessard, Julie, Jiang I. Wu, Jeffrey A. Ranish, Mimi Wan, Monte M. Winslow, Brett T. Staahl, Hai Wu, Ruedi Aebersold, Isabella A. Graef, and Gerald R. Crabtree. 2007. "An

- Essential Switch in Subunit Composition of a Chromatin Remodeling Complex during Neural Development." *Neuron* 55 (2): 201–15.
- Liao, Yang, Gordon K. Smyth, and Wei Shi. 2014. "featureCounts: An Efficient General Purpose Program for Assigning Sequence Reads to Genomic Features." *Bioinformatics* 30 (7): 923–30.
- Ling, Hui, Riccardo Spizzo, Yaser Atlasi, Milena Nicoloso, Masayoshi Shimizu, Roxana S. Redis, Naohiro Nishida, et al. 2013. "CCAT2, a Novel Noncoding RNA Mapping to 8q24, Underlies Metastatic Progression and Chromosomal Instability in Colon Cancer." *Genome Research* 23 (9): 1446–61.
- Liu, Guang-Hui, Basam Z. Barkho, Sergio Ruiz, Dinh Diep, Jing Qu, Sheng-Lian Yang, Athanasia D. Panopoulos, et al. 2011. "Recapitulation of Premature Ageing with iPSCs from Hutchinson-Gilford Progeria Syndrome." *Nature* 472 (7342): 221–25.
- Liu, Jimmy Z., Suzanne van Sommeren, Hailiang Huang, Siew C. Ng, Rudi Alberts, Atsushi Takahashi, Stephan Ripke, et al. 2015. "Association Analyses Identify 38 Susceptibility Loci for Inflammatory Bowel Disease and Highlight Shared Genetic Risk across Populations." *Nature Genetics* 47 (9): 979–86.
- Liu, Wenjun, Hailong Wu, Lili Chen, Yankai Wen, Xiaoni Kong, and Wei-Qiang Gao. 2015. "Park7 Interacts with p47(phox) to Direct NADPH Oxidase-Dependent ROS Production and Protect against Sepsis." *Cell Research* 25 (6): 691–706.
- Li, Yang I., Bryce van de Geijn, Anil Raj, David A. Knowles, Allegra A. Petti, David Golan, Yoav Gilad, and Jonathan K. Pritchard. 2016. "RNA Splicing Is a Primary Link between Genetic Variation and Disease." *Science* 352 (6285): 600–604.
- Li, Yang I., David A. Knowles, and Jonathan K. Pritchard. 2016. "LeafCutter: Annotation-Free Quantification of RNA Splicing." *bioRxiv*. doi:10.1101/044107.
- Li, Yue, and Manolis Kellis. 2016. "RIVIERA-MT: A Bayesian Model to Infer Risk Variants in Related Traits Using Summary Statistics and Functional Genomic Annotations." *bioRxiv*. doi:10.1101/059345.
- Love, Michael I., Wolfgang Huber, and Simon Anders. 2014. "Moderated Estimation of Fold Change and Dispersion for RNA-Seq Data with DESeq2." doi:10.1101/002832.
- Lu, Qiongshi, Xinwei Yao, Yiming Hu, and Hongyu Zhao. 2016. "GenoWAP: GWAS Signal Prioritization through Integrated Analysis of Genomic Functional Annotation." *Bioinformatics* 32 (4): 542–48.
- Manolio, Teri A., Francis S. Collins, Nancy J. Cox, David B. Goldstein, Lucia A. Hindorff, David J. Hunter, Mark I. McCarthy, et al. 2009. "Finding the Missing Heritability of Complex Diseases." *Nature* 461 (7265): 747–53.
- Mathelier, Anthony, Beibei Xin, Tsu-Pei Chiu, Lin Yang, Remo Rohs, and Wyeth W. Wasserman. 2016. "DNA Shape Features Improve Transcription Factor Binding Site Predictions In Vivo." *Cell Systems* 3 (3): 278–86.e4.
- McCarthy, Davis J., Peter Humburg, Alexander Kanapin, Manuel A. Rivas, Kyle Gaulton, Jean-Baptiste Cazier, Peter Donnelly, and asds. 2014. "Choice of Transcripts and Software Has a Large Effect on Variant Annotation." *Genome Medicine* 6 (3): 26.
- McCarthy, Shane, Sayantan Das, Warren Kretzschmar, Olivier Delaneau, Andrew R. Wood, Alexander Teumer, Hyun Min Kang, et al. 2016. "A Reference Panel of 64,976 Haplotypes for Genotype Imputation." *Nature Genetics* 48 (10): 1279–83.
- McClellan, Jon, and Mary-Claire King. 2010. "Genetic Heterogeneity in Human Disease." *Cell* 141 (2): 210–17.
- McLaren, William, Laurent Gil, Sarah E. Hunt, Harpreet Singh Riat, Graham R. S. Ritchie, Anja Thormann, Paul Flliceck, and Fiona Cunningham. 2016. "The Ensembl Variant Effect Predictor." *Genome Biology* 17 (1): 122.

- Melchionda, Laura, Tobias B. Haack, Steven Hardy, Truus E. M. Abbink, Erika Fernandez-Vizarra, Eleonora Lamantea, Silvia Marchet, et al. 2014. "Mutations in APOPT1, Encoding a Mitochondrial Protein, Cause Cavitating Leukoencephalopathy with Cytochrome c Oxidase Deficiency." *American Journal of Human Genetics* 95 (3): 315–25.
- Mele, M., P. G. Ferreira, F. Reverter, D. S. DeLuca, J. Monlong, M. Sammeth, T. R. Young, et al. 2015. "The Human Transcriptome across Tissues and Individuals." *Science* 348 (6235): 660–65.
- Melzer, David, John R. B. Perry, Dena Hernandez, Anna-Maria Corsi, Kara Stevens, Ian Rafferty, Fulvio Lauretani, et al. 2008. "A Genome-Wide Association Study Identifies Protein Quantitative Trait Loci (pQTLs)." *PLoS Genetics* 4 (5): e1000072.
- Moyerbrailean, Gregory A., Cynthia A. Kalita, Chris T. Harvey, Xiaoquan Wen, Francesca Luca, and Roger Pique-Regi. 2016. "Which Genetics Variants in DNase-Seq Footprints Are More Likely to Alter Binding?" *PLoS Genetics* 12 (2): e1005875.
- Mumbach, Maxwell R., Ansuman T. Satpathy, Evan A. Boyle, Chao Dai, Benjamin G. Gowen, Seung Woo Cho, Michelle L. Nguyen, et al. 2017. "Enhancer Connectome in Primary Human Cells Identifies Target Genes of Disease-Associated DNA Elements." *Nature Genetics*, September. doi:10.1038/ng.3963.
- Musunuru, Kiran, Alanna Strong, Maria Frank-Kamenetsky, Noemi E. Lee, Tim Ahfeldt, Katherine V. Sachs, Xiaoyu Li, et al. 2010. "From Noncoding Variant to Phenotype via SORT1 at the 1p13 Cholesterol Locus." *Nature* 466 (7307): 714–19.
- Newman, Aaron M., Chih Long Liu, Michael R. Green, Andrew J. Gentles, Weiguo Feng, Yue Xu, Chuong D. Hoang, Maximilian Diehn, and Ash A. Alizadeh. 2015. "Robust Enumeration of Cell Subsets from Tissue Expression Profiles." *Nature Methods* 12 (5): 453–57.
- Nica, Alexandra C., Leopold Parts, Daniel Glass, James Nisbet, Amy Barrett, Magdalena Sekowska, Mary Travers, et al. 2011. "The Architecture of Gene Regulatory Variation across Multiple Human Tissues: The MuTHER Study." *PLoS Genetics* 7 (2): e1002003.
- Nicolae, Dan L., Eric Gamazon, Wei Zhang, Shiwei Duan, M. Eileen Dolan, and Nancy J. Cox. 2010. "Trait-Associated SNPs Are More Likely to Be eQTLs: Annotation to Enhance Discovery from GWAS." *PLoS Genetics* 6 (4): e1000888.
- Okada, Yukinori, Di Wu, Gosia Trynka, Towfique Raj, Chikashi Terao, Katsunori Ikari, Yuta Kochi, et al. 2014. "Genetics of Rheumatoid Arthritis Contributes to Biology and Drug Discovery." *Nature* 506 (7488): 376–81.
- Okbay, Aysu, Jonathan P. Beauchamp, Mark Alan Fontana, James J. Lee, Tune H. Pers, Cornelius A. Rietveld, Patrick Turley, et al. 2016. "Genome-Wide Association Study Identifies 74 Loci Associated with Educational Attainment." *Nature* 533 (7604): 539–42.
- Ongen, Halit, Alfonso Buil, Andrew Anand Brown, Emmanouil T. Dermitzakis, and Olivier Delaneau. 2016. "Fast and Efficient QTL Mapper for Thousands of Molecular Phenotypes." *Bioinformatics* 32 (10): 1479–85.
- Palazzo, Alexander F., and Eliza S. Lee. 2015. "Non-Coding RNA: What Is Functional and What Is Junk?" *Frontiers in Genetics* 6 (January): 2.
- Panopoulos, Athanasia D., Matteo D'Antonio, Paola Benaglio, Roy Williams, Sherin I. Hashem, Bernhard M. Schuldt, Christopher DeBoever, et al. 2017. "iPSCORE: A Resource of 222 iPSC Lines Enabling Functional Characterization of Genetic Variation across a Variety of Cell Types." *Stem Cell Reports*, April. doi:10.1016/j.stemcr.2017.03.012.
- Papalex, Efthymia, and Rahul Satija. 2017. "Single-Cell RNA Sequencing to Explore Immune Cell Heterogeneity." *Nature Reviews Immunology*, August. doi:10.1038/nri.2017.76.

- Parkes, Miles, Adrian Cortes, David A. van Heel, and Matthew A. Brown. 2013. "Genetic Insights into Common Pathways and Complex Relationships among Immune-Mediated Diseases." *Nature Reviews Genetics* 14 (9): 661–73.
- Pashos, Evangelia E., Yoson Park, Xiao Wang, Avanthi Raghavan, Wenli Yang, Deepa Abbey, Derek T. Peters, et al. 2017. "Large, Diverse Population Cohorts of hiPSCs and Derived Hepatocyte-like Cells Reveal Functional Genetic Variation at Blood Lipid-Associated Loci." *Cell Stem Cell* 20 (4): 558–70.e10.
- Pasquali, Lorenzo, Kyle J. Gaulton, Santiago A. Rodríguez-Seguí, Loris Mularoni, Irene Miguel-Escalada, İldem Akerman, Juan J. Tena, et al. 2014. "Pancreatic Islet Enhancer Clusters Enriched in Type 2 Diabetes Risk-Associated Variants." *Nature Genetics* 46 (2): 136–43.
- Passier, Robert, Valeria Orlova, and Christine Mummery. 2016. "Complex Tissue and Disease Modeling Using hiPSCs." *Cell Stem Cell* 18 (3): 309–21.
- Peltkova, Vanya D., Richard F. Wintle, Laurence A. Rubin, Christopher I. Amos, Qiqing Huang, Xiangjun Gu, Bill Newman, et al. 2004. "Functional Variants of OCTN Cation Transporter Genes Are Associated with Crohn Disease." *Nature Genetics* 36 (5): 471–75.
- Pers, Tune H., Pascal Timshel, and Joel N. Hirschhorn. 2015. "SNPsnap: A Web-Based Tool for Identification and Annotation of Matched SNPs." *Bioinformatics* 31 (3): 418–20.
- Peters, Marjolein J., Linda Broer, Hanneke L. D. M. Willemen, Gudny Eiriksdottir, Lynne J. Hocking, Kate L. Holliday, Michael A. Horan, et al. 2013. "Genome-Wide Association Study Meta-Analysis of Chronic Widespread Pain: Evidence for Involvement of the 5p15.2 Region." *Annals of the Rheumatic Diseases* 72 (3): 427–36.
- Pickrell, Joseph K. 2014. "Joint Analysis of Functional Genomic Data and Genome-Wide Association Studies of 18 Human Traits." *American Journal of Human Genetics* 94 (4): 559–73.
- Pickrell, Joseph K., John C. Marioni, Athma A. Pai, Jacob F. Degner, Barbara E. Engelhardt, Everlyne Nkadori, Jean-Baptiste Veyrieras, Matthew Stephens, Yoav Gilad, and Jonathan K. Pritchard. 2010. "Understanding Mechanisms Underlying Human Gene Expression Variation with RNA Sequencing." *Nature* 464 (7289): 768–72.
- Polo, Jose M., Susanna Liu, Maria Eugenia Figueroa, Warakorn Kulalert, Sarah Eminli, Kah Yong Tan, Effie Apostolou, et al. 2010. "Cell Type of Origin Influences the Molecular and Functional Properties of Mouse Induced Pluripotent Stem Cells." *Nature Biotechnology* 28 (8): 848–55.
- Praetorius, Christian, Christine Grill, Simon N. Stacey, Alexander M. Metcalf, David U. Gorkin, Kathleen C. Robinson, Eric Van Otterloo, et al. 2013. "A Polymorphism in IRF4 Affects Human Pigmentation through a Tyrosinase-Dependent MITF/TFAP2A Pathway." *Cell* 155 (5): 1022–33.
- Raghavan, Avanthi, Raghavan Avanthi, Wang Xiao, Rogov Peter, Wang Li, Zhang Xiaolan, Tarjei S. Mikkelsen, and Musunuru Kiran. 2016. "High-Throughput Screening and CRISPR-Cas9 Modeling of Causal Lipid-Associated Expression Quantitative Trait Locus Variants." doi:10.1101/056820.
- Ramasamy, Adaikalavan, Daniah Trabzuni, Sebastian Guelfi, Vibin Varghese, Colin Smith, Robert Walker, Tisham De, et al. 2014. "Genetic Variability in the Regulation of Gene Expression in Ten Regions of the Human Brain." *Nature Neuroscience* 17 (10): 1418–28.
- Rao, Suhas S. P., Miriam H. Huntley, Neva C. Durand, Elena K. Stamenova, Ivan D. Bochkov, James T. Robinson, Adrian L. Sanborn, et al. 2014. "A 3D Map of the Human Genome at Kilobase Resolution Reveals Principles of Chromatin Looping." *Cell* 159 (7): 1665–80.
- Ritchie, Graham R. S., Ian Dunham, Eleftheria Zeggini, and Paul Flückeck. 2014. "Functional

- Annotation of Noncoding Sequence Variants." *Nature Methods* 11 (3): 294–96.
- Roadmap Epigenomics Consortium, Anshul Kundaje, Wouter Meuleman, Jason Ernst, Misha Bilenky, Angela Yen, Alireza Heravi-Moussavi, et al. 2015. "Integrative Analysis of 111 Reference Human Epigenomes." *Nature* 518 (7539): 317–30.
- Robinton, Daisy A., and George Q. Daley. 2012. "The Promise of Induced Pluripotent Stem Cells in Research and Therapy." *Nature* 481 (7381): 295–305.
- Ruffell, Daniela, Foteini Mourkioti, Adriana Gambardella, Peggy Kirstetter, Rodolphe G. Lopez, Nadia Rosenthal, and Claus Nerlov. 2009. "A CREB-C/EBPbeta Cascade Induces M2 Macrophage-Specific Gene Expression and Promotes Muscle Injury Repair." *Proceedings of the National Academy of Sciences of the United States of America* 106 (41): 17475–80.
- Ryan, Niamh M., Stewart W. Morris, David J. Porteous, Martin S. Taylor, and Kathryn L. Evans. 2014. "SuRFing the Genomics Wave: An R Package for Prioritising SNPs by Functionality." *Genome Medicine* 6 (10): 79.
- Sala, Luca, Milena Bellin, and Christine L. Mummery. 2016. "Integrating Cardiomyocytes from Human Pluripotent Stem Cells in Safety Pharmacology: Has the Time Come?" *British Journal of Pharmacology*, September. doi:10.1111/bph.13577.
- Schizophrenia Working Group of the Psychiatric Genomics Consortium. 2014. "Biological Insights from 108 Schizophrenia-Associated Genetic Loci." *Nature* 511 (7510): 421–27.
- Schork, Andrew J., Wesley K. Thompson, Phillip Pham, Ali Torkamani, J. Cooper Roddey, Patrick F. Sullivan, John R. Kelsoe, et al. 2013. "All SNPs Are Not Created Equal: Genome-Wide Association Studies Reveal a Consistent Pattern of Enrichment among Functionally Annotated SNPs." *PLoS Genetics* 9 (4): e1003449.
- Sheffield, Nathan C., and Bock Christoph. 2015. "LOLA: Enrichment Analysis for Genomic Region Sets and Regulatory Elements in R and Bioconductor." *Bioinformatics* 32 (4): 587–89.
- Sheffield, Nathan C., Robert E. Thurman, Lingyun Song, Alexias Safi, John A. Stamatoyannopoulos, Boris Lenhard, Gregory E. Crawford, and Terrence S. Furey. 2013. "Patterns of Regulatory Activity across Diverse Human Cell Types Predict Tissue Identity, Transcription Factor Binding, and Long-Range Interactions." *Genome Research* 23 (5): 777–88.
- Shihab, Hashem A., Mark F. Rogers, Julian Gough, Matthew Mort, David N. Cooper, Ian N. M. Day, Tom R. Gaunt, and Colin Campbell. 2015. "An Integrative Approach to Predicting the Functional Effects of Non-Coding and Coding Sequence Variation." *Bioinformatics* 31 (10): 1536–43.
- Siepel, Adam, Gill Bejerano, Jakob S. Pedersen, Angie S. Hinrichs, Minmei Hou, Kate Rosenbloom, Hiram Clawson, et al. 2005. "Evolutionarily Conserved Elements in Vertebrate, Insect, Worm, and Yeast Genomes." *Genome Research* 15 (8): 1034–50.
- Singh, Tarjinder, Adam P. Levine, Philip J. Smith, Andrew M. Smith, Anthony W. Segal, and Jeffrey C. Barrett. 2015. "Characterization of Expression Quantitative Trait Loci in the Human Colon." *Inflammatory Bowel Diseases* 21 (2): 251–56.
- Smemo, Scott, Juan J. Tena, Kyoung-Han Kim, Eric R. Gamazon, Noboru J. Sakabe, Carlos Gómez-Marín, Ivy Aneas, et al. 2014. "Obesity-Associated Variants within FTO Form Long-Range Functional Connections with IRX3." *Nature* 507 (7492): 371–75.
- Smith, Brenden W., Sarah S. Rozelle, Amy Leung, Jessalyn Ubellacker, Ashley Parks, Shirley K. Nah, Deborah French, et al. 2013. "The Aryl Hydrocarbon Receptor Directs Hematopoietic Progenitor Cell Expansion and Differentiation." *Blood* 122 (3): 376–85.
- Soldner, Frank, Yonatan Stelzer, Chikdu S. Shivalila, Brian J. Abraham, Jeanne C. Latourelle, M. Inmaculada Barrasa, Johanna Goldmann, Richard H. Myers, Richard A. Young, and Rudolf Jaenisch. 2016. "Parkinson-Associated Risk Variant in Distal Enhancer of α -

- Synuclein Modulates Target Gene Expression." *Nature* 533 (7601): 95–99.
- Spain, Sarah L., and Jeffrey C. Barrett. 2015. "Strategies for Fine-Mapping Complex Traits." *Human Molecular Genetics* 24 (R1): R111–19.
- Speed, Doug, Na Cai, UCLEB Consortium, Michael R. Johnson, Sergey Nejentsev, and David J. Balding. 2017. "Reevaluation of SNP Heritability in Complex Human Traits." *Nature Genetics* 49 (7): 986–92.
- Spilker, Christina, Spilker Christina, and Michael R. Kreutz. 2010. "RapGAPs in Brain: Multipurpose Players in Neuronal Rap Signalling." *The European Journal of Neuroscience* 32 (1): 1–9.
- Spitz, François. 2016. "Gene Regulation at a Distance: From Remote Enhancers to 3D Regulatory Ensembles." *Seminars in Cell & Developmental Biology* 57: 57–67.
- Stark, Amy L., Ronald J. Hause Jr, Lidiya K. Gorsic, Nirav N. Antao, Shan S. Wong, Sophie H. Chung, Daniel F. Gill, et al. 2014. "Protein Quantitative Trait Loci Identify Novel Candidates Modulating Cellular Response to Chemotherapy." *PLoS Genetics* 10 (4): e1004192.
- Stenson, Peter D., Matthew Mort, Edward V. Ball, Katy Shaw, Andrew Phillips, and David N. Cooper. 2014. "The Human Gene Mutation Database: Building a Comprehensive Mutation Repository for Clinical and Molecular Genetics, Diagnostic Testing and Personalized Genomic Medicine." *Human Genetics* 133 (1): 1–9.
- Sudmant, Peter H., Tobias Rausch, Eugene J. Gardner, Robert E. Handsaker, Alexej Abyzov, John Huddleston, Yan Zhang, et al. 2015. "An Integrated Map of Structural Variation in 2,504 Human Genomes." *Nature* 526 (7571): 75–81.
- Takahashi, Kazutoshi, and Shinya Yamanaka. 2006. "Induction of Pluripotent Stem Cells from Mouse Embryonic and Adult Fibroblast Cultures by Defined Factors." *Cell* 126 (4): 663–76.
- Tan, Minjia, Hao Luo, Sangkyu Lee, Fulai Jin, Jeong Soo Yang, Emilie Montellier, Thierry Buchou, et al. 2011. "Identification of 67 Histone Marks and Histone Lysine Crotonylation as a New Type of Histone Modification." *Cell* 146 (6): 1016–28.
- Teng, Mingxiang, Michael I. Love, Carrie A. Davis, Sarah Djebali, Alexander Dobin, Brenton R. Graveley, Sheng Li, et al. 2016. "A Benchmark for RNA-Seq Quantification Pipelines." *Genome Biology* 17 (April): 74.
- Tewhey, Ryan, Dylan Kotliar, Daniel S. Park, Brandon Liu, Sarah Winnicki, Steven K. Reilly, Kristian G. Andersen, et al. 2016. "Direct Identification of Hundreds of Expression-Modulating Variants Using a Multiplexed Reporter Assay." *Cell* 165 (6): 1519–29.
- Thurner, Matthias, Martijn van de Bunt, Kyle Gaulton, Amy Barrett, Amanda J. Bennett, Jason M. Torres, Vibe Nylander, et al. 2017. "Integration of Human Pancreatic Islet Genomic Data Refines Regulatory Mechanisms at Type 2 Diabetes Susceptibility Loci." doi:10.1101/190892.
- Trynka, Gosia, Cynthia Sandor, Buhm Han, Han Xu, Barbara E. Stranger, X. Shirley Liu, and Soumya Raychaudhuri. 2013. "Chromatin Marks Identify Critical Cell Types for Fine Mapping Complex Trait Variants." *Nature Genetics* 45 (2): 124–30.
- Trynka, Gosia, Harm-Jan Westra, Kamil Slowikowski, Xinli Hu, Han Xu, Barbara E. Stranger, Robert J. Klein, Buhm Han, and Soumya Raychaudhuri. 2015. "Disentangling the Effects of Colocalizing Genomic Annotations to Functionally Prioritize Non-Coding Variants within Complex-Trait Loci." *American Journal of Human Genetics* 97 (1): 139–52.
- Turner, Adam W., Amy Martinuk, Anada Silva, Paulina Lau, Majid Nikpay, Per Eriksson, Lasse Folkersen, et al. 2016. "Functional Analysis of a Novel Genome-Wide Association Study Signal in SMAD3 That Confers Protection From Coronary Artery Disease." *Arteriosclerosis, Thrombosis, and Vascular Biology* 36 (5): 972–83.

- Vaquerizas, Juan M., Sarah K. Kummerfeld, Sarah A. Teichmann, and Nicholas M. Luscombe. 2009. "A Census of Human Transcription Factors: Function, Expression and Evolution." *Nature Reviews. Genetics* 10 (4): 252–63.
- Veerman, Christiaan C., Georgios Kosmidis, Christine L. Mummery, Simona Casini, Arie O. Verkerk, and Milena Bellin. 2015. "Immaturity of Human Stem-Cell-Derived Cardiomyocytes in Culture: Fatal Flaw or Soluble Problem?" *Stem Cells and Development* 24 (9): 1035–52.
- Wainger, Brian J., Evangelos Kiskinis, Cassidy Mellin, Ole Wiskow, Steve S. W. Han, Jackson Sandoe, Numa P. Perez, et al. 2014. "Intrinsic Membrane Hyperexcitability of Amyotrophic Lateral Sclerosis Patient-Derived Motor Neurons." *Cell Reports* 7 (1): 1–11.
- Wakefield, Jon. 2009. "Bayes Factors for Genome-Wide Association Studies: Comparison with P-Values." *Genetic Epidemiology* 33 (1): 79–86.
- Wallace, Chris, Antony J. Cutler, Nikolas Pontikos, Marcin L. Pekalski, Oliver S. Burren, Jason D. Cooper, Arcadio Rubio García, et al. 2015. "Dissection of a Complex Disease Susceptibility Region Using a Bayesian Stochastic Search Approach to Fine Mapping." *PLoS Genetics* 11 (6): e1005272.
- Wang, Xinchen, Liang He, Sarah Goggin, Alham Saadat, Li Wang, Melina Claussnitzer, and Manolis Kellis. 2017. "High-Resolution Genome-Wide Functional Dissection of Transcriptional Regulatory Regions in Human." *bioRxiv*. doi:10.1101/193136.
- Ward, Lucas D., and Manolis Kellis. 2012. "HaploReg: A Resource for Exploring Chromatin States, Conservation, and Regulatory Motif Alterations within Sets of Genetically Linked Variants." *Nucleic Acids Research* 40 (Database issue): D930–34.
- Warren, Curtis R., Cashell E. Jaquish, and Chad A. Cowan. 2017. "The NextGen Genetic Association Studies Consortium: A Foray into In Vitro Population Genetics." *Cell Stem Cell* 20 (4): 431–33.
- Warren, Curtis R., John F. O'Sullivan, Max Friesen, Caroline E. Becker, Xiaoling Zhang, Poching Liu, Yoshiyuki Wakabayashi, et al. 2017. "Induced Pluripotent Stem Cell Differentiation Enables Functional Validation of GWAS Variants in Metabolic Disease." *Cell Stem Cell* 20 (4): 547–57.e7.
- Wellcome Trust Case Control Consortium. 2007. "Genome-Wide Association Study of 14,000 Cases of Seven Common Diseases and 3,000 Shared Controls." *Nature* 447 (7145): 661–78.
- Wellcome Trust Case Control Consortium, Julian B. Maller, Gilean McVean, Jake Byrnes, Damjan Vukcevic, Kimmo Palin, Zhan Su, et al. 2012. "Bayesian Refinement of Association Signals for 14 Loci in 3 Common Diseases." *Nature Genetics* 44 (12): 1294–1301.
- Wernig, Marius, Jian-Ping Zhao, Jan Pruszak, Eva Hedlund, Dongdong Fu, Frank Soldner, Vania Broccoli, Martha Constantine-Paton, Ole Isacson, and Rudolf Jaenisch. 2008. "Neurons Derived from Reprogrammed Fibroblasts Functionally Integrate into the Fetal Brain and Improve Symptoms of Rats with Parkinson's Disease." *Proceedings of the National Academy of Sciences of the United States of America* 105 (15): 5856–61.
- Westra, Harm-Jan, Marjolein J. Peters, Tõnu Esko, Hanieh Yaghootkar, Claudia Schurmann, Johannes Kettunen, Mark W. Christiansen, et al. 2013. "Systematic Identification of Trans eQTLs as Putative Drivers of Known Disease Associations." *Nature Genetics* 45 (10): 1238–43.
- Willis, Dianna E., Meng Wang, Elizabeth Brown, Lilah Fones, and John W. Cave. 2016. "Selective Repression of Gene Expression in Neuropathic Pain by the Neuron-Restrictive Silencing Factor/repressor Element-1 Silencing Transcription (NRSF/REST)." *Neuroscience Letters* 625 (June): 20–25.
- Yang, Jian, Andrew Bakshi, Zhihong Zhu, Gibran Hemani, Anna A. E. Vinkhuyzen, Sang

- Hong Lee, Matthew R. Robinson, et al. 2015. "Genetic Variance Estimation with Imputed Variants Finds Negligible Missing Heritability for Human Height and Body Mass Index." *Nature Genetics* 47 (10): 1114–20.
- Young, Gareth T., Gutteridge Alex, Heather DE Fox, Anna L. Wilbrey, Cao Lishuang, Lily T. Cho, Adam R. Brown, et al. 2014. "Characterizing Human Stem Cell-derived Sensory Neurons at the Single-Cell Level Reveals Their Ion Channel Expression and Utility in Pain Research." *Molecular Therapy: The Journal of the American Society of Gene Therapy* 22 (8): 1530–43.
- Zhang, Xiaoyang, Richard Cowper-Sal Iari, Swneke D. Bailey, Jason H. Moore, and Mathieu Lupien. 2012. "Integrative Functional Genomics Identifies an Enhancer Looping to the SOX9 Gene Disrupted by the 17q24.3 Prostate Cancer Risk Locus." *Genome Research* 22 (8): 1437–46.
- Zhao, Hao, Zhifu Sun, Jing Wang, Haojie Huang, Jean-Pierre Kocher, and Liguo Wang. 2014. "CrossMap: A Versatile Tool for Coordinate Conversion between Genome Assemblies." *Bioinformatics* 30 (7): 1006–7.
- Zhou, Jian, and Olga G. Troyanskaya. 2015. "Predicting Effects of Noncoding Variants with Deep Learning-based Sequence Model." *Nature Methods* 12 (10): 931–34.
- Zhou, Pingzhu, Aibin He, and William T. Pu. 2012. "Regulation of GATA4 Transcriptional Activity in Cardiovascular Development and Disease." *Current Topics in Developmental Biology* 100: 143–69.
- Zhu, Zhihong, Futao Zhang, Han Hu, Andrew Bakshi, Matthew R. Robinson, Joseph E. Powell, Grant W. Montgomery, et al. 2016. "Integration of Summary Data from GWAS and eQTL Studies Predicts Complex Trait Gene Targets." *Nature Genetics* 48 (5): 481–87.