Bibliography

1000 Genomes Project Consortium, Adam Auton, Lisa D Brooks, Richard M Durbin, Erik P Garrison, Hyun Min Kang, Jan O Korbel, Jonathan L Marchini, Shane McCarthy, et al. (2015). "A global reference for human genetic variation". *Nature* 526.7571, pp. 68–74.

Abecasis, Gonçalo R, David Altshuler, Adam Auton, Lisa D Brooks, Richard M Durbin, Richard A Gibbs, Matt E Hurles, and Gil A McVean (2010). "A map of human genome variation from population-scale sequencing". *Nature* 467.7319, pp. 1061–1073.

Albert, Frank W and Leonid Kruglyak (2015). "The role of regulatory variation in complex traits and disease". *Nat. Rev. Genet.* 16.4, pp. 197–212.

Altshuler, David M, Richard A Gibbs, Leena Peltonen, Emmanouil Dermitzakis, Stephen F Schaffner, Fuli Yu, Penelope E Bonnen, Paul I W de Bakker, Panos Deloukas, et al. (2010). "Integrating common and rare genetic variation in diverse human populations". *Nature* 467.7311, pp. 52–58.

Anderson, Carl A, Fredrik H Pettersson, Geraldine M Clarke, Lon R Cardon, Andrew P Morris, and Krina T Zondervan (2010). "Data quality control in genetic case-control association studies". *Nat. Protoc.* 5.9, pp. 1564–1573.

Anderson, Carl A, Gabrielle Boucher, Charlie W Lees, Andre Franke, Mauro D'Amato, Kent D Taylor, James C Lee, Philippe Goyette, Marcin Imielinski, et al. (2011). "Meta-analysis identifies 29 additional ulcerative colitis risk loci, increasing the number of confirmed associations to 47". *Nat. Genet.* 43.3, pp. 246–252.

Andersson, Robin, Claudia Gebhard, Irene Miguel-Escalada, Ilka Hoof, Jette Bornholdt, Mette Boyd, Yun Chen, Xiaobei Zhao, Christian Schmidl, et al. (2014). "An atlas of active enhancers across human cell types and tissues". *Nature* 507, pp. 455–461.

Antonopoulos, Dionysios A, Susan M Huse, Hilary G Morrison, Thomas M Schmidt, Mitchell L Sogin, and Vincent B Young (2009). "Reproducible community dynamics of the gastrointestinal microbiota following antibiotic perturbation". *Infect. Immun.* 77.6, pp. 2367–2375.

Asano, Kouichi, Tomonaga Matsushita, Junji Umeno, Naoya Hosono, Atsushi Takahashi, Takahisa Kawaguchi, Takayuki Matsumoto, Toshiyuki Matsui, Yoichi Kakuta, et al. (2009). "A genome-wide association study identifies three new susceptibility loci for ulcerative colitis in the Japanese population". *Nat. Genet.* 41.12, pp. 1325–1329.

Asimit, Jennifer L, Aaron G Day-Williams, Andrew P Morris, and Eleftheria Zeggini (2012). "ARIEL and AMELIA: testing for an accumulation of rare variants using next-generation sequencing data". *Hum. Hered.* 73.2, pp. 84–94.

Barbieri, Christopher E, Sylvan C Baca, Michael S Lawrence, Francesca Demichelis, Mirjam Blattner, Jean-Philippe Theurillat, Thomas A White, Petar Stojanov, Eliezer Van Allen, et al. (2012). "Exome sequencing identifies recurrent SPOP, FOXA1 and MED12 mutations in prostate cancer". *Nat. Genet.* 44.6, pp. 685–689.

Barrett, Jeffrey C and Lon R Cardon (2006). "Evaluating coverage of genome-wide association studies". *Nat. Genet.* 38.6, pp. 659–662.

Barrett, Jeffrey C, Sarah Hansoul, Dan L Nicolae, Judy H Cho, Richard H Duerr, John D Rioux, Steven R Brant, Mark S Silverberg, Kent D Taylor, et al. (2008). "Genome-wide association defines more than 30 distinct susceptibility loci for Crohn's disease". *Nat. Genet.* 40.8, pp. 955–962.

Barrett, Jeffrey C, James C Lee, Charles W Lees, Natalie J Prescott, Carl A Anderson, Anne Phillips, Emma Wesley, Kirstie Parnell, Hu Zhang, et al. (2009). "Genome-wide association study of ulcerative colitis identifies three new susceptibility loci, including the HNF4A region". *Nat. Genet.* 41.12, pp. 1330–1334.

Battle, Alexis, Sara Mostafavi, Xiaowei Zhu, James B Potash, Myrna M Weissman, Courtney McCormick, Christian D Haudenschild, Kenneth B Beckman, Jianxin Shi, et al. (2014). "Characterizing the genetic basis of transcriptome diversity through RNA-sequencing of 922 individuals". *Genome Res.* 24.1, pp. 14–24.

Baumgart, D C and W J Sandborn (2007). "Inflammatory bowel disease: clinical aspects and established and evolving therapies". *Lancet* 369.5, pp. 1641–1657.

Beaudoin, Mélissa, Philippe Goyette, Gabrielle Boucher, Ken Sin Lo, Manuel A Rivas, Christine Stevens, Azadeh Alikashani, Martin Ladouceur, David Ellinghaus, et al. (2013). "Deep Resequencing of GWAS Loci Identifies Rare Variants in CARD9, IL23R and RNF186 That Are Associated with Ulcerative Colitis". *PLoS Genet.* 9.9.

Belton, Jon-Matthew, Rachel Patton McCord, Johan Harmen Gibcus, Natalia Naumova, Ye Zhan, and Job Dekker (2012). "Hi-C: a comprehensive technique to capture the conformation of genomes". *Methods* 58.3, pp. 268–276.

Bernstein, Charles N, Michael Fried, J H Krabshuis, Henry Cohen, R Eliakim, Suleiman Fedail, Richard Gearry, K L Goh, Saheed Hamid, et al. (2010). "World gastroenterology organization practice guidelines for the diagnosis and management of IBD in 2010". *Inflamm. Bowel Dis.* 16.1, pp. 112–124.

Blaydon, Diana C, Paolo Biancheri, Wei-Li Di, Vincent Plagnol, Rita M Cabral, Matthew A Brooke, David A van Heel, Franz Ruschendorf, Mark Toynbee, et al. (2011). "Inflammatory skin and bowel disease linked to ADAM17 deletion". *N. Engl. J. Med.* 365.16, pp. 1502–1508.

Botstein, D, R L White, M Skolnick, and R W Davis (1980). "Construction of a genetic linkage map in man using restriction fragment length polymorphisms". *Am. J. Hum. Genet.* 32.3, pp. 314–331.

Boyd, Andy, Jean Golding, John Macleod, Debbie A Lawlor, Abigail Fraser, John Henderson, Lynn Molloy, Andy Ness, Susan Ring, and George Davey Smith (2013). "Cohort Profile: the 'children of the 90s'–the index offspring of the Avon Longitudinal Study of Parents and Children". *Int. J. Epidemiol.* 42.1, pp. 111–127.

Boyman, Onur and Jonathan Sprent (2012). "The role of interleukin-2 during homeostasis and activation of the immune system". *Nat. Rev. Immunol.* 12.3, pp. 180–190.

Brant, Steven R (2011). "Update on the heritability of inflammatory bowel disease: the importance of twin studies". *Inflamm. Bowel Dis.* 17.1, pp. 1–5.

Brant, Steven R, Yifan Fu, Carter T Fields, Romulo Baltazar, Geoffrey Ravenhill, Michael R Pickles, Patrick M Rohal, Jasdeep Mann, Barbara S Kirschner, et al. (1998). "American families with Crohn's disease have strong evidence for linkage to chromosome 16 but not chromosome 12". *Gastroenterology* 115, pp. 1056–1061.

Brown, Eric M, Manish Sadarangani, and B Brett Finlay (2013). "The role of the immune system in governing host-microbe interactions in the intestine". *Nat. Immunol.* 14.7, pp. 660–667.

Browning, Brian L and Sharon R Browning (2016). "Genotype Imputation with Millions of Reference Samples". *Am. J. Hum. Genet.* 98.1, pp. 116–126.

Bulik-Sullivan, Brendan K, Po-Ru Loh, Hilary K Finucane, Stephan Ripke, Jian Yang, Schizophrenia Working Group of the Psychiatric Genomics Consortium, Nick Patterson, Mark J Daly, Alkes L Price, and Benjamin M Neale (2015). "LD Score regression distinguishes confounding from polygenicity in genome-wide association studies". *Nat. Genet.* 47.3, pp. 291–295.

Cai, Na, Tim B Bigdeli, Warren Kretzschmar, Yihan Li, Jieqin Liang, Li Song, Jingchu Hu, Qibin Li, Wei Jin, et al. (2015). "Sparse whole-genome sequencing identifies two loci for major depressive disorder". *Nature* 12415800.

Carson, Kenneth R, Daniele Focosi, Eugene O Major, Mario Petrini, Elizabeth A Richey, Dennis P West, and Charles L Bennett (2009). "Monoclonal antibody-associated progressive multifocal leucoencephalopathy in patients treated with rituximab, natalizumab, and efalizumab: a Review from the Research on Adverse Drug Events and Reports (RADAR) Project". *Lancet Oncol.* 10.8, pp. 816–824.

Cavanaugh, J A, D F Callen, S R Wilson, P M Stanford, M E Sraml, M Gorska, J Crawford, S A Whitmore, C Shlegel, et al. (1998). "Analysis of Australian Crohn's disease pedigrees refines the localization for susceptibility to inflammatory bowel disease on chromosome 16". *Ann. Hum. Genet.* 62.4, pp. 291–298.

Cavanaugh, J and The International IBD Genetics Consortium (2001). "International Collaboration Provides Convincing Linkage Replication in Complex Disease through Analysis of a Large Pooled Data Set : Crohn Disease and Chromosome 16". *Am. J. Hum. Genet.* 68, pp. 1165–1171.

Chang, Christopher C, Carson C Chow, Laurent Cam Tellier, Shashaank Vattikuti, Shaun M Purcell, and James J Lee (2015). "Second-generation PLINK: rising to the challenge of larger and richer datasets". *Gigascience* 4.1, p. 7.

Chang, Diana, Feng Gao, Andrea Slavney, Li Ma, Yedael Y Waldman, Aaron J Sams, Paul Billing-Ross, Aviv Madar, Richard Spritz, and Alon Keinan (2014). "Accounting for eXentricities: Analysis of the X Chromosome in GWAS Reveals X-Linked Genes Implicated in Autoimmune Diseases". *PLoS One* 9.12, e113684.

Chen, Gui-Bo, Sang Hong Lee, Marie-Jo A Brion, Grant W Montgomery, Naomi R Wray, Graham L Radford-Smith, Peter M Visscher, and International IBD Genetics Consortium (2014). "Estimation and partitioning of (co)heritability of inflammatory bowel disease from GWAS and immunochip data". *Hum. Mol. Genet.* 23.17, pp. 4710–4720.

Choi, Murim, Ute I Scholl, Weizhen Ji, Tiewen Liu, Irina R Tikhonova, Paul Zumbo, Ahmet Nayir, Ayin Bakkaloğlu, Seza Özen, et al. (2009). "Genetic diagnosis by whole exome capture and massively parallel DNA sequencing". *Proceedings of the National Academy of Sciences* 106.45, pp. 19096–19101.

Cleynen, Isabelle, Juan R González, Carolina Figueroa, Andre Franke, Dermot McGovern, Martin Bortlík, Bart J A Crusius, Maurizio Vecchi, Marta Artieda, et al. (2013). "Genetic factors conferring an increased susceptibility to develop Crohn's disease also influence disease phenotype: results from the IBDchip European Project". *Gut* 62.11, pp. 1556–1565.

Cleynen, Isabelle, Gabrielle Boucher, Luke Jostins, L Philip Schumm, Sebastian Zeissig, Tariq Ahmad, Vibeke Andersen, Jane M Andrews, Vito Annese, et al. (2016). "Inherited determinants of Crohn's disease and

ulcerative colitis phenotypes: a genetic association study". *Lancet* 387.10014, pp. 156–167.

Colombel, Jean-Frédéric, William J Sandborn, Paul Rutgeerts, Robert Enns, Stephen B Hanauer, Remo Panaccione, Stefan Schreiber, Dan Byczkowski, Ju Li, et al. (2007). "Adalimumab for maintenance of clinical response and remission in patients with Crohn's disease: the CHARM trial". *Gastroenterology* 132.1, pp. 52–65.

Curran, Mark E, K I T F Lau, Jochen Hampe, Stefan Schreiber, Steven Bridger, Andrew J S Macpherson, L O N R Cardon, Hakan Sakul, Timothy J R Harris, et al. (1998). "Genetic Analysis of Inflammatory Bowel Disease in a Large European Cohort Supports Linkage to Chromosomes 12 and 16". *Gastroenterology* 115, pp. 1066–1071.

Dahle, Maria K, Anders E Myhre, Ansgar O Aasen, and Jacob E Wang (2005). "Effects of forskolin on Kupffer cell production of interleukin-10 and tumor necrosis factor alpha differ from those of endogenous adenylyl cyclase activators: possible role for adenylyl cyclase 9". *Infect. Immun.* 73.11, pp. 7290–7296.

Danjou, Fabrice, Magdalena Zoledziewska, Carlo Sidore, Maristella Steri, Fabio Busonero, Andrea Maschio, Antonella Mulas, Lucia Perseu, Susanna Barella, et al. (2015). "Genome-wide association analyses based on whole-genome sequencing in Sardinia provide insights into regulation of hemoglobin levels". *Nat. Genet.* 47.11, pp. 1264–1271.

Davey Smith, George and Gibran Hemani (2014). "Mendelian randomization: genetic anchors for causal inference in epidemiological studies". *Hum. Mol. Genet.* 23.R1, R89–98.

Daye, Z John, Hongzhe Li, and Zhi Wei (2012). "A powerful test for multiple rare variants association studies that incorporates sequencing qualities". *Nucleic Acids Res.* 40.8, e60.

DePristo, Mark A, Eric Banks, Ryan Poplin, Kiran V Garimella, Jared R Maguire, Christopher Hartl, Anthony A Philippakis, Guillermo del Angel, Manuel A Rivas, et al. (2011). "A framework for variation discovery and genotyping using next-generation DNA sequencing data". *Nat. Genet.* 43.5, pp. 491–498.

Dendrou, Calliope A, Adrian Cortes, Lydia Shipman, Hayley G Evans, Kathrine E Attfield, Luke Jostins, Thomas Barber, Gurman Kaur, Subita Balaram Kuttikkatte, et al. (2016). "Resolving TYK2 locus genotype-to-phenotype differences in autoimmunity". *Sci. Transl. Med.* 8.363, 363ra149–363ra149.

Derkach, Andriy, Jerry F Lawless, and Lei Sun (2013). "Robust and Powerful Tests for Rare Variants Using Fisher's Method to Combine Evidence of Association From Two or More Complementary Tests". *Genet. Epidemiol.* 37.1, pp. 110–121.

Derkach, Andriy, Theodore Chiang, Jiafen Gong, Laura Addis, Sara Dobbins, Ian Tomlinson, Richard Houlston, Deb K Pal, and Lisa J Strug (2014). "Association analysis using next-generation sequence data from publicly available control groups: The robust variance score statistic". *Bioinformatics* 30.15, pp. 2179–2188.

Dethlefsen, Les, Sue Huse, Mitchell L Sogin, and David A Relman (2008). "The pervasive effects of an antibiotic on the human gut microbiota, as revealed by deep 16s rRNA sequencing". *PLoS Biol.* 6.11, pp. 2383–2400.

de Vries, A Boudewijn, Marcel Janse, Hans Blokzijl, and Rinse K Weersma (2015). "Distinctive inflammatory bowel disease phenotype in primary sclerosing cholangitis". *World J. Gastroenterol.* 21.6, pp. 1956–1971.

D'haeseleer, Patrik (2006). "What are DNA sequence motifs?" *Nat. Biotechnol.* 24.4, pp. 423–425.

Dilthey, Alexander T, Pierre-Antoine Gourraud, Alexander J Mentzer, Nezih Cereb, Zamin Iqbal, and Gil McVean (2016). "High-Accuracy HLA Type Inference from Whole-Genome Sequencing Data Using Population Reference Graphs". *PLoS Comput. Biol.* 12.10, e1005151.

Dilthey, Alexander, Charles Cox, Zamin Iqbal, Matthew R Nelson, and Gil McVean (2015). "Improved genome inference in the MHC using a population reference graph". *Nat. Genet.* 47.6, pp. 682–688.

Duan, Biyan, Richard Davis, Eva L Sadat, Julie Collins, Paul C Sternweis, Dorothy Yuan, and Lily I Jiang (2010). "Distinct roles of adenylyl cyclase VII in regulating the immune responses in mice". *J. Immunol.* 185.1, pp. 335–344.

Duerr, R H, K D Taylor, S R Brant, J D Rioux, M S Silverberg, M J Daly, a H Steinhart, C Abraham, M Regueiro, et al. (2006). "A Genome-Wide Association Study Identifies IL23R as an Inflammatory Bowel Disease Gene". *Science* 314.5804, pp. 1461–1463.

Durbin, Richard (2014). "Efficient haplotype matching and storage using the positional Burrows-Wheeler transform (PBWT)". *Bioinformatics* 30.9, pp. 1266–1272.

ENCODE Project Consortium (2012). "An integrated encyclopedia of DNA elements in the human genome". *Nature* 489.7414, pp. 57–74.

Eiseman, B, W Silen, G S Bascom, and A J Kauvar (1958). "Fecal enema as an adjunct in the treatment of pseudomembranous enterocolitis". *Surgery* 44.5, pp. 854–859.

Ellinghaus, David, Luke Jostins, Sarah L Spain, Adrian Cortes, Jörn Bethune, Buhm Han, Yu Rang Park, Soumya Raychaudhuri, Jennie G Pouget, et al. (2016). "Analysis of five chronic inflammatory diseases identifies 27 new associations and highlights disease-specific patterns at shared loci". *Nat. Genet.* 48.5, pp. 510–518.

Evans, David M and Lon R Cardon (2004). "Guidelines for genotyping in genomewide linkage studies: single-nucleotide-polymorphism maps versus microsatellite maps". *Am. J. Hum. Genet.* 75.4, pp. 687–692.

Fairfax, Benjamin P, Seiko Makino, Jayachandran Radhakrishnan, Katharine Plant, Stephen Leslie, Alexander Dilthey, Peter Ellis, Cordelia Langford, Fredrik O Vannberg, and Julian C Knight (2012). "Genetics of gene expression in primary immune cells identifies cell type-specific master regulators and roles of HLA alleles". *Nat. Genet.* 44.5, pp. 502–510.

Fairfax, Benjamin P, Peter Humburg, Seiko Makino, Vivek Naranbhai, Daniel Wong, Evelyn Lau, Luke Jostins, Katharine Plant, Robert Andrews, et al. (2014). "Innate immune activity conditions the effect of regulatory variants upon monocyte gene expression". *Science* 343.6175, p. 1246949.

Farh, Kyle Kai-How, Alexander Marson, Jiang Zhu, Markus Kleinewietfeld, William J Housley, Samantha Beik, Noam Shoresh, Holly Whitton, Russell J H Ryan, et al. (2015). "Genetic and epigenetic fine mapping of causal autoimmune disease variants". *Nature* 518.7539, pp. 337–343.

Feagan, Brian G, Paul Rutgeerts, Bruce E Sands, Stephen Hanauer, Jean-Frédéric Colombel, William J Sandborn, Gert Van Assche, Jeffrey Axler, Hyo-Jong Kim, et al. (2013). "Vedolizumab as induction and maintenance therapy for ulcerative colitis". *N. Engl. J. Med.* 369.8, pp. 699–710.

Fisher, Sheila A, Mark Tremelling, Carl A Anderson, Rhian Gwilliam, Suzannah Bumpstead, Natalie J Prescott, Elaine R Nimmo, Dunecan Massey, Carlo Berzuini, et al. (2008). "Genetic determinants of ulcerative colitis include the ECM1 locus and five loci implicated in Crohn's disease". *Nat. Genet.* 40.6, pp. 710–712.

Ford, D, D F Easton, M Stratton, S Narod, D Goldgar, P Devilee, D T Bishop, B Weber, G Lenoir, et al. (1998). "Genetic Heterogeneity and Penetrance Analysis of the BRCA1 and BRCA2 Genes in Breast Cancer Families". *Am. J. Hum. Genet.* 62.3, pp. 676–689.

Franke, Andre, Tobias Balschun, Tom H Karlsen, Jurgita Sventoraityte, Susanna Nikolaus, Gabriele Mayr, Francisco S Domingues, Mario Albrecht, Michael Nothnagel, et al. (2008). "Sequence variants in IL10, ARPC2 and multiple other loci contribute to ulcerative colitis susceptibility". *Nat. Genet.* 40.11, pp. 1319–1323.

Franke, Andre, Dermot P B McGovern, Jeffrey C Barrett, Kai Wang, Graham L Radford-Smith, Tariq Ahmad, Charlie W Lees, Tobias Balschun, James Lee, et al. (2010). "Genome-wide meta-analysis increases to 71 the number of confirmed Crohn's disease susceptibility loci". *Nat. Genet.* 42.12, pp. 1118–1125.

Fu, Guoping, Yuhong Chen, James Schuman, Demin Wang, and Renren Wen (2012). "Phospholipase Cγ2 plays a role in TCR signal transduction and T cell selection". *J. Immunol.* 189.5, pp. 2326–2332.

Fuchsberger, Christian, Jason Flannick, Tanya M Teslovich, Anubha Mahajan, Vineeta Agarwala, Kyle J Gaulton, Clement Ma, Pierre Fontanillas, Loukas Moutsianas, et al. (2016). "The genetic architecture of type 2 diabetes". *Nature* 536.7614, pp. 41–47.

GTEx Consortium (2015). "Human genomics. The Genotype-Tissue Expression (GTEx) pilot analysis: multitissue gene regulation in humans". *Science* 348.6235, pp. 648–660.

Ganna, Andrea, Giulio Genovese, Daniel P Howrigan, Andrea Byrnes, Mitja I Kurki, Seyedeh M Zekavat, Christopher W Whelan, Mart Kals, Michel G Nivard, et al. (2016). "Ultra-rare disruptive and damaging mutations influence educational attainment in the general population". *Nat. Neurosci.* 19.12, pp. 1563–1565.

Garner, Chad (2011). "Confounded by sequencing depth in association studies of rare alleles". *Genet. Epidemiol.* 35.4, pp. 261–268.

Genovese, Giulio, Menachem Fromer, Eli A Stahl, Douglas M Ruderfer, Kimberly Chambert, Mikael Landén, Jennifer L Moran, Shaun M Purcell, Pamela Sklar, et al. (2016). "Increased burden of ultra-rare protein-altering variants among 4,877 individuals with schizophrenia". *Nat. Neurosci.* 19.11, pp. 1433–1441.

Gevers, Dirk, Subra Kugathasan, Lee A Denson, Yoshiki Vázquez-Baeza, Will Van Treuren, Boyu Ren, Emma Schwager, Dan Knights, Se Jin Song, et al. (2014). "The treatment-naive microbiome in new-onset Crohn's disease". *Cell Host Microbe* 15.3, pp. 382–392.

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 Genet.* 10.5, e1004383.

Gibson, Greg (2011). "Rare and common variants: twenty arguments". *Nat. Rev. Genet.* 13.2, pp. 135–145.

Glocker, E O, D Kotlarz, Kaan Boztug, E Michael Gertz, Alejandro A Schäffer, Fatih Noyan, Mario Perro, Jana Diestelhorst, Anna Allroth, et al. (2009). "Inflammatory Bowel Disease and Mutations Affecting the Interleukin-10 Receptor". *N. Engl. J. Med.* 361.21, pp. 2033–2045.

Goodwin, Sara, John D McPherson, and W Richard McCombie (2016). "Coming of age: ten years of next-generation sequencing technologies". *Nat. Rev. Genet.* 17.6, pp. 333–351.

Goyette, Philippe, Gabrielle Boucher, Dermot Mallon, Eva Ellinghaus, Luke Jostins, Hailiang Huang, Stephan Ripke, Elena S Gusareva, Vito Annese, et al. (2015). "High-density mapping of the MHC identifies a shared role for HLA-DRB1*01:03 in inflammatory bowel diseases and heterozygous advantage in ulcerative colitis". *Nat. Genet.* 47.2, pp. 172–179.

Gusella, J F, N S Wexler, P M Conneally, S L Naylor, M a Anderson, R E Tanzi, P C Watkins, K Ottina, M R Wallace, et al. (1983). "A polymorphic DNA marker genetically linked to Huntington's disease". *Nature* 306.17, pp. 234–238.

Hall, Stephen S (2013). "Genetics: a gene of rare effect". *Nature* 496.7444, pp. 152–155.

Halme, Leena, P Paavola-Sakki, Ulla Turunen, Maarit Lappalainen, Martti Farkkila, and Kimmo Kontula (2006). "Family and twin studies in inflammatory bowel disease". *World J. Gastroenterol.* 12.23, pp. 3668–3672.

Hampe, Jochen, Andre Franke, Philip Rosenstiel, Andreas Till, Markus Teuber, Klaus Huse, Mario Albrecht, Gabriele Mayr, Francisco M De La Vega, et al. (2007). "A genome-wide association scan of nonsynonymous SNPs identifies a susceptibility variant for Crohn disease in ATG16L1". *Nat. Genet.* 39.2, pp. 207–211.

Han, Jian-Wen, Hou-Feng Zheng, Yong Cui, Liang-Dan Sun, Dong-Qing Ye, Zhi Hu, Jin-Hua Xu, Zhi-Ming Cai, Wei Huang, et al. (2009). "Genome-wide association study in a Chinese Han population identifies nine new susceptibility loci for systemic lupus erythematosus". *Nat. Genet.* 41.11, pp. 1234–1237.

Hanauer, Stephen B, Brian G Feagan, Gary R Lichtenstein, Lloyd F Mayer, S Schreiber, Jean Frederic Colombel, Daniel Rachmilewitz, Douglas C Wolf, Allan Olson, et al. (2002). "Maintenance infliximab for Crohn's disease: the ACCENT I randomised trial". *Lancet* 359.9317, pp. 1541–1549.

Handsaker, Robert E, Vanessa Van Doren, Jennifer R Berman, Giulio Genovese, Seva Kashin, Linda M Boettger, and Steven A McCarroll (2015). "Large multiallelic copy number variations in humans". *Nat. Genet.* 47.3, pp. 296–303.

Haritunians, Talin, Kent D Taylor, Stephan R Targan, Maria Dubinsky, Andrew Ippoliti, Soonil Kwon, Xiuqing Guo, Gil Y Melmed, Dror Berel, et al. (2010). "Genetic Predictors of Medically Refractory Ulcerative Colitis". *Inflamm. Bowel Dis.* 16.11, pp. 1830–1840.

Hosten, Terron Anthony, Ke Zhao, Hong Qiu Han, Gang Liu, and Xiang Hui He (2014). "Alicaforsen: An Emerging Therapeutic Agent for Ulcerative Colitis and Refractory Pouchitis". *Gastroenterol. Res. Pract.* 7.2, pp. 51–55.

Hu, Yi-Juan, Peizhou Liao, H Richard Johnston, Andrew S Allen, and Glen A Satten (2016). "Testing Rare-Variant Association without Calling Genotypes Allows for Systematic Differences in Sequencing between Cases and Controls". *PLoS Genet.* 12.5, e1006040.

Huang, Hailiang, Ming Fang, Luke Jostins, Masa U Mirkov, Gabrielle Boucher, Carl A Anderson, Vibeke Andersen, Isabelle Cleynen, Adrian Cortes, et al. (2015). "Association mapping of inflammatory bowel disease loci to single variant resolution". *bioRxiv*, p. 028688.

Hugot, J P, P Laurent-Puig, C Gower-Rousseau, J M Olson, J C Lee, L Beaugerie, I Naom, J L Dupas, A Van Gossum, et al. (1996). "Mapping of a susceptibility locus for Crohn's disease on chromosome 16". *Nature* 379.6568, pp. 821–823.

Hugot, Jean-Pierre, Mathias Chamaillard, Habib Zouali, Suzanne Lesage, Jean-Pierre Cézard, Jacques Belaiche, Sven Almer, Curt Tysk, Colm A O'Morain, et al. (2001). "Association of NOD2 leucine-rich repeat variants with susceptibility to Crohn's disease". *Nature* 411.6837, pp. 599–603.

Hunt, Karen a, Vanisha Mistry, Nicholas a Bockett, Tariq Ahmad, Maria Ban, Jonathan N Barker, Jeffrey C Barrett, Hannah Blackburn, Oliver Brand, et al. (2013). "Negligible impact of rare autoimmune-locus coding-region variants on missing heritability". *Nature* 498.7453, pp. 232–235.

Hynes, Richard O (2002). "Integrins: bidirectional, allosteric signaling machines". *Cell* 110.6, pp. 673–687.

Ioannidis, John P A (2003). *Genetic associations: False or true?*

Ivanov, Ivaylo I, Brent S McKenzie, Liang Zhou, Carlos E Tadokoro, Alice Lepelley, Juan J Lafaille, Daniel J Cua, and Dan R Littman (2006). "The Orphan Nuclear Receptor RORγt Directs the Differentiation Program of Proinflammatory IL-17+ T Helper Cells". *Cell* 126.6, pp. 1121–1133.

James, Dustin G, Da Hea Seo, Jiajing Chen, Caroline Vemulapalli, and Christian D Stone (2011). "Efalizumab, a human monoclonal anti-CD11a antibody, in the treatment of moderate to severe Crohn's disease: An open-label pilot study". *Dig. Dis. Sci.* 56.6, pp. 1806–1810.

Jiang, Lily I, Paul C Sternweis, and Jennifer E Wang (2013). "Zymosan activates protein kinase A via adenylyl cyclase VII to modulate innate immune responses during inflammation". *Mol. Immunol.* 54.1, pp. 14–22.

Jin, Jill (2014). "JAMA patient page. Inflammatory bowel disease". *JAMA* 311.19, p. 2034.

Jostins, Luke, Stephan Ripke, Rinse K Weersma, Richard H Duerr, Dermot P McGovern, Ken Y Hui, James C Lee, L Philip Schumm, Yashoda Sharma, et al. (2012). "Host-microbe interactions have shaped the genetic architecture of inflammatory bowel disease". *Nature* 491.7422, pp. 119–124.

Julià, Antonio, Eugeni Domènech, María Chaparro, Valle García-Sánchez, Fernando Gomollón, Julián Panés, Míriam Mañosa, Manuel Barreiro-De Acosta, Ana Gutiérrez, et al. (2014). "A genome-wide association study identifies a novel

locus at 6q22.1 associated with ulcerative colitis". *Hum. Mol. Genet.* 23.25, pp. 6927–6934.

Juyal, Garima, Sapna Negi, Ajit Sood, Aditi Gupta, Pushplata Prasad, Sabyasachi Senapati, Jacques Zaneveld, Shalini Singh, Vandana Midha, et al. (2015). "Genome-wide association scan in north Indians reveals three novel HLA-independent risk loci for ulcerative colitis". *Gut* 64, pp. 571–579.

Kandt, R S, J L Haines, M Smith, H Northrup, R J Gardner, M P Short, K Dumars, E S Roach, S Steingold, and S Wall (1992). "Linkage of an important gene locus for tuberous sclerosis to a chromosome 16 marker for polycystic kidney disease". *Nat. Genet.* 2.1, pp. 37–41.

Kaplan, Gilaad G (2015). "The global burden of IBD: from 2015 to 2025". *Nat. Rev. Gastroenterol. Hepatol.* 12.12, pp. 720–727.

Kenny, Eimear E, Itsik Pe'er, Amir Karban, Laurie Ozelius, Adele A Mitchell, Sok Meng Ng, Monica Erazo, Harry Ostrer, Clara Abraham, et al. (2012). "A genome-wide scan of Ashkenazi Jewish Crohn's disease suggests novel susceptibility loci". *PLoS Genet.* 8.3, e1002559.

Kheradpour, Pouya and Manolis Kellis (2014). "Systematic discovery and characterization of regulatory motifs in ENCODE TF binding experiments". *Nucleic Acids Res.* 42.5, pp. 2976–2987.

Khor, Bernard, Agnès Gardet, and Ramnik J Xavier (2011). "Genetics and pathogenesis of inflammatory bowel disease". *Nature* 474, pp. 308–317.

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 Genet.* 10.10, e1004722.

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". *Nat. Genet.* 46.3, pp. 310–315.

Korneliussen, Thorfinn, Anders Albrechtsen, and Rasmus Nielsen (2014). "ANGSD: Analysis of Next Generation Sequencing Data". *BMC Bioinformatics* 15, p. 356.

Kostic, Aleksandar D, Ramnik J Xavier, and Dirk Gevers (2014). "The microbiome in inflammatory bowel disease: current status and the future ahead". *Gastroenterology* 146.6, pp. 1489–1499.

Kunkel, Eric J and Eugene C Butcher (2003). "Plasma-cell homing". *Nat. Rev. Immunol.* 3.10, pp. 822–829.

Lee, James C, Daniele Biasci, Rebecca Roberts, Richard B Gearry, John C Mansfield, Tariq Ahmad, Natalie J Prescott, Jack Satsangi, David C Wilson, et al. (2017). "Genome-wide association study identifies

distinct genetic contributions to prognosis and susceptibility in Crohn's disease". *Nat. Genet.* 49.2, pp. 262–268.

Lee, Mark N, Chun Ye, Alexandra-Chloé Villani, Towfique Raj, Weibo Li, Thomas M Eisenhaure, Selina H Imboywa, Portia I Chipendo, F Ann Ran, et al. (2014a). "Common genetic variants modulate pathogen-sensing responses in human dendritic cells". *Science* 343.6175, p. 1246980.

Lee, Phil H, Colm O'Dushlaine, Brett Thomas, and Shaun M Purcell (2012a). "INRICH: interval-based enrichment analysis for genome-wide association studies". *Bioinformatics* 28.13, pp. 1797–1799.

Lee, Seunggeun, Michael C Wu, and Xihong Lin (2012b). "Optimal tests for rare variant effects in sequencing association studies". *Biostatistics* 13.4, pp. 762–775.

Lee, Seunggeung, Gonçalo R Abecasis, Michael Boehnke, and Xihong Lin (2014b). "Rare-variant association analysis: study designs and statistical tests". *Am. J. Hum. Genet.* 95.1, pp. 5–23.

Leek, Jeffrey T, Robert B Scharpf, Héctor Corrada Bravo, David Simcha, Benjamin Langmead, W Evan Johnson, Donald Geman, Keith Baggerly, and Rafael A Irizarry (2010). "Tackling the widespread and critical impact of batch effects in high-throughput data". *Nat. Rev. Genet.* 11.10, pp. 733–739.

Lek, Monkol, Konrad J Karczewski, Eric V Minikel, Kaitlin E Samocha, Eric Banks, Timothy Fennell, Anne H ODonnell-Luria, James S Ware, Andrew J Hill, et al. (2016). "Analysis of protein-coding genetic variation in 60,706 humans". *Nature* 536.7616, pp. 285–291.

Li, Bingshan and S M Leal (2008). "Methods for detecting associations with rare variants for common diseases: application to analysis of sequence data". *Am. J. Hum. Genet.* 83, pp. 311–321.

Li, Bingshan, Dajiang J Liu, and Suzanne M Leal (2013a). "Identifying rare variants associated with complex traits via sequencing". *Curr. Protoc. Hum. Genet.* Pp. 1.26.1–1.26.22.

Li, Heng (2011). "A statistical framework for SNP calling, mutation discovery, association mapping and population genetical parameter estimation from sequencing data". *Bioinformatics* 27.21, pp. 2987–2993.

Li, Heng, Bob Handsaker, Alec Wysoker, Tim Fennell, Jue Ruan, Nils Homer, Gabor Marth, Goncalo Abecasis, Richard Durbin, and 1000 Genome Project Data Processing Subgroup (2009). "The Sequence Alignment/Map format and SAMtools". *Bioinformatics* 25.16, pp. 2078–2079.

Li, Heng, Bob Handsaker, Petr Danecek, Shane McCarthy, and John Marshall (2013b). *SAMtools and BCFtools*. URL: `https://sourceforge.net/projects/samtools/files/samtools/0.1.19/`.

Li, Yun R, Jin Li, Sihai D Zhao, Jonathan P Bradfield, Frank D Mentch, S Melkorka Maggadottir, Cuiping Hou, Debra J Abrams, Diana Chang, et al.

(2015). "Meta-analysis of shared genetic architecture across ten pediatric autoimmune diseases". *Nat. Med.* 21.9, pp. 1018–1027.

Li, Yun, Carlo Sidore, Hyun Min Kang, Micheal Boehnke, and Gonçalo R Abecasis (2011). "Low-coverage sequencing: implications for design of complex trait association studies". *Genome Res.* 21, pp. 940–951.

Libioulle, Cécile, Edouard Louis, Sarah Hansoul, Cynthia Sandor, Frédéric Farnir, Denis Franchimont, Séverine Vermeire, Olivier Dewit, Martine De Vos, et al. (2007). "Novel Crohn disease locus identified by genome-wide association maps to a gene desert on 5p13.1 and modulates expression of PTGER4". *PLoS Genet.* 3.4, pp. 0538–0543.

Liu, Jimmy Z and Carl A Anderson (2014). "Genetic studies of Crohn's disease: past, present and future". *Best Pract. Res. Clin. Gastroenterol.* 28.3, pp. 373–386.

Liu, Jimmy Z, Suzanne van Sommeren, Hailiang Huang, Siew C Ng, Rudi Alberts, Atsushi Takahashi, Stephan Ripke, James C Lee, Luke Jostins, et al. (2015). "Association analyses identify 38 susceptibility loci for inflammatory bowel disease and highlight shared genetic risk across populations". *Nat. Genet.* 47.9, pp. 979–989.

Loftus, Edward V (2004). "Clinical epidemiology of inflammatory bowel disease: incidence, prevalence, and environmental influences". *Gastroenterology* 126.6, pp. 1504–1517.

Louis, Thomas A (1982). "Finding the Observed Information Matrix when Using the EM Algorithm". *J. R. Stat. Soc. Series B Stat. Methodol.* 44.2, pp. 226–233.

Luci, Carmelo, Ana Reynders, Ivaylo I Ivanov, Celine Cognet, Laurent Chiche, Lionel Chasson, Jean Hardwigsen, Esperanza Anguiano, Jacques Banchereau, et al. (2009). "Influence of the transcription factor RORγt on the development of NKp46+ cell populations in gut and skin". *Nat. Immunol.* 10.1, pp. 75–82.

Luo, Yang, Katrina M de Lange, Luke Jostins, Loukas Moutsianas, Joshua Randall, Nicholas A Kennedy, Christopher A Lamb, Shane McCarthy, Tariq Ahmad, et al. (2017). "Exploring the genetic architecture of inflammatory bowel disease by whole-genome sequencing identifies association at ADCY7". *Nat. Genet.* 49.2, pp. 186–192.

M'Koma, Amosy E (2013). "Inflammatory bowel disease: an expanding global health problem". *Clin. Med. Insights Gastroenterol.* 6, pp. 33–47.

Madsen, Bo Eskerod and Sharon R Browning (2009). "A groupwise association test for rare mutations using a weighted sum statistic". *PLoS Genet.* 5.2, e1000384.

Maher, Brendan (2008). "The case of the missing heritability". *Nature News Features* 456, pp. 18–21.

Manichaikul, Ani, Josyf C Mychaleckyj, Stephen S Rich, Kathy Daly, Michèle Sale, and Wei-Min Chen (2010). "Robust relationship inference in genome-wide association studies". *Bioinformatics* 26.22, pp. 2867–2873.

Manolio, Teri A, Francis S Collins, Nancy J Cox, David B Goldstein, Lucia A Hindorff, David J Hunter, Mark I McCarthy, Erin M Ramos, Lon R Cardon, et al. (2009). "Finding the missing heritability of complex diseases". *Nature* 461.7265, pp. 747–753.

Marceau, Kristine, Minni T B McMaster, Taylor F Smith, Joost G Daams, Catharina E M van Beijsterveldt, Dorret I Boomsma, and Valerie S Knopik (2016). "The Prenatal Environment in Twin Studies: A Review on Chorionicity". *Behav. Genet.* 46.3, pp. 286–303.

Marchini, Jonathan and Bryan Howie (2010). "Genotype imputation for genome-wide association studies". *Nat. Rev. Genet.* 11.7, pp. 499–511.

Matute, Juan D, Andres A Arias, Nicola A M Wright, Iwona Wrobel, Christopher C M Waterhouse, Xing Jun Li, Christophe C Marchal, Natalie D Stull, David B Lewis, et al. (2009). "A new genetic subgroup of chronic granulomatous disease with autosomal recessive mutations in p40 phox and selective defects in neutrophil NADPH oxidase activity". *Blood* 114.15, pp. 3309–3315.

Maurano, Matthew T, Richard Humbert, Eric Rynes, Robert E Thurman, Eric Haugen, Hao Wang, Alex P Reynolds, Richard Sandstrom, Hongzhu Qu, et al. (2012). "Systematic localization of common disease-associated variation in regulatory DNA". *Science* 337.6099, pp. 1190–1195.

McCarthy, Shane, Sayantan Das, Warren Kretzschmar, Olivier Delaneau, Andrew R Wood, Alexander Teumer, Hyun Min Kang, Christian Fuchsberger, Petr Danecek, et al. (2016). "A reference panel of 64,976 haplotypes for genotype imputation". *Nat. Genet.* 48.10, pp. 1279–1283.

McLaren, William, Bethan Pritchard, Daniel Rios, Yuan Chen, Paul Flicek, and Fiona Cunningham (2010). "Deriving the consequences of genomic variants with the Ensembl API and SNP Effect Predictor". *Bioinformatics* 26.16, pp. 2069–2070.

McVean, Gilean A T, Simon R Myers, Sarah Hunt, Panos Deloukas, David R Bentley, and Peter Donnelly (2004). "The fine-scale structure of recombination rate variation in the human genome". *Science* 304, pp. 581–584.

Meynert, Alison M, Morad Ansari, David R FitzPatrick, and Martin S Taylor (2014). "Variant detection sensitivity and biases in whole genome and exome sequencing". *BMC Bioinformatics* 15, p. 247.

Miceli-Richard, C, S Lesage, M Rybojad, A M Prieur, S Manouvrier-Hanu, R Häfner, M Chamaillard, H Zouali, G Thomas, and J P Hugot (2001). "CARD15 mutations in Blau syndrome". *Nat. Genet.* 29.1, pp. 19–20.

Mifsud, Borbala, Filipe Tavares-Cadete, Alice N Young, Robert Sugar, Stefan Schoenfelder, Lauren Ferreira, Steven W Wingett, Simon Andrews, William Grey, et al. (2015). "Mapping long-range promoter contacts in human cells with high-resolution capture Hi-C". *Nat. Genet.* 47.6, pp. 598–606.

Mills, Ryan E, W Stephen Pittard, Julienne M Mullaney, Umar Farooq, Todd H Creasy, Anup A Mahurkar, David M Kemeza, Daniel S Strassler, Chris P Ponting, et al. (2011). "Natural genetic variation caused by small insertions and deletions in the human genome". *Genome Res.* 21.6, pp. 830–839.

Moayyeri, Alireza, Christopher J Hammond, Deborah J Hart, and Timothy D Spector (2013). "The UK Adult Twin Registry (TwinsUK Resource)". *Twin Res. Hum. Genet.* 16.1, pp. 144–149.

Molodecky, Natalie A, Ing Shian Soon, Doreen M Rabi, William A Ghali, Mollie Ferris, Greg Chernoff, Eric I Benchimol, Remo Panaccione, Subrata Ghosh, et al. (2012). "Increasing incidence and prevalence of the inflammatory bowel diseases with time, based on systematic review". *Gastroenterology* 142.1, pp. 46–54.

Monteleone, Giovanni, Markus F Neurath, Sandro Ardizzone, Antonio Di Sabatino, Massimo C Fantini, Fabiana Castiglione, Maria L Scribano, Alessandro Armuzzi, Flavio Caprioli, et al. (2015). "Mongersen, an oral SMAD7 antisense oligonucleotide, and Crohn's disease". *N. Engl. J. Med.* 372.12, pp. 1104–1113.

Morgenthaler, Stephan and William G Thilly (2007). "A strategy to discover genes that carry multi-allelic or mono-allelic risk for common diseases: a cohort allelic sums test (CAST)". *Mutat. Res.* 615.1-2, pp. 28–56.

Morris, Andrew P and Eleftheria Zeggini (2010). "An evaluation of statistical approaches to rare variant analysis in genetic association studies". *Genet. Epidemiol.* 34.2, pp. 188–193.

Moutsianas, Loukas and Andrew P Morris (2014). "Methodology for the analysis of rare genetic variation in genome-wide association and re-sequencing studies of complex human traits". *Brief. Funct. Genomics* 13.5, pp. 362–370.

Neale, Benjamin M, Manuel a Rivas, Benjamin F Voight, David Altshuler, Bernie Devlin, Marju Orho-Melander, Sekar Kathiresan, Shaun M Purcell, Kathryn Roeder, and Mark J Daly (2011). "Testing for an unusual distribution of rare variants". *PLoS Genet.* 7.3, e1001322.

Negoro, K, D P B McGovern, Y Kinouchi, S Takahashi, N J Lench, T Shimosegawa, A Carey, L R Cardon, D P Jewell, and D A Van Heel (2003). "Analysis of the IBD5 locus and potential gene-gene interactions in Crohns disease". *Gut* 52, pp. 541–546.

Nejentsev, Sergey, Joanna M M Howson, Neil M Walker, Jeffrey Szeszko, Sarah F Field, Helen E Stevens, Pamela Reynolds, Matthew Hardy, Erna King,

et al. (2007). "Localization of type 1 diabetes susceptibility to the MHC class I genes HLA-B and HLA-A". *Nature* 450.7171, pp. 887–892.

Ng, Sarah B, Emily H Turner, Peggy D Robertson, Steven D Flygare, Abigail W Bigham, Choli Lee, Tristan Shaffer, Michelle Wong, Arindam Bhattacharjee, et al. (2009). "Targeted Capture and Massively Parallel Sequencing of Twelve Human Exomes". *Nature* 461.7261, pp. 272–276.

Ng, Sarah B, Kati J Buckingham, Choli Lee, Abigail W Bigham, Holly K Tabor, Karin M Dent, Chad D Huff, Paul T Shannon, Ethylin Wang Jabs, et al. (2010). "Exome sequencing identifies the cause of a mendelian disorder". *Nat. Genet.* 42.1, pp. 30–35.

Novak, Adam M, Glenn Hickey, Erik Garrison, Sean Blum, Abram Connelly, Alexander Dilthey, Jordan Eizenga, M A Saleh Elmohamed, Sally Guthrie, et al. (2017). "Genome Graphs".

Ogura, Yasunori, Denise K Bonen, Naohiro Inohara, Dan L Nicolae, Felicia F Chen, Richard Ramos, Heidi Britton, Thomas Moran, Reda Karaliuskas, et al. (2001). "A frameshift mutation in NOD2 associated with susceptibility to Crohn's disease". *Nature* 411.6837, pp. 603–606.

Ohmen, Jeffrey D, Hui-Ying Yang, Karen K Yamamoto, Hong-Yu Zhao, Yuanhong Ma, L Gordon Bentley, Zhihan Huang, Scott Gerwehr, Sheila Pressman, et al. (1996). "Susceptibility locus for inflammatory bowel disease on chromosome 16 has a role in Crohns disease, but not in ulcerative colitis". *Hum. Mol. Genet.* 5.10, pp. 1679–1683.

Ombrello, Michael J, Elaine F Remmers, Guangping Sun, Alexandra F Freeman, Shrimati Datta, Parizad Torabi-Parizi, Naeha Subramanian, Tom D Bunney, Rhona W Baxendale, et al. (2012). "Cold urticaria, immunodeficiency, and autoimmunity related to PLCG2 deletions". *N. Engl. J. Med.* 366.4, pp. 330–338.

Pals, Steven T, David J J de Gorter, and Marcel Spaargaren (2007). "Lymphoma dissemination: the other face of lymphocyte homing". *Blood* 110.9, pp. 3102–3111.

Pan, Wei (2009). "Asymptotic tests of association with multiple SNPs in linkage disequilibrium". *Genet. Epidemiol.* 33.6, pp. 497–507.

Parkes, M, J Satsangi, G M Lathrop, J I Bell, and D P Jewell (1996). "Susceptibility loci in inflammatory bowel disease". *Lancet* 348.9041, p. 1588.

Parkes, Miles, Jeffrey C Barrett, Natalie J Prescott, Mark Tremelling, Carl A Anderson, Sheila A Fisher, Roland G Roberts, Elaine R Nimmo, Fraser R Cummings, et al. (2007). "Sequence variants in the autophagy gene IRGM and multiple other replicating loci contribute to Crohn's disease susceptibility". *Nat. Genet.* 39.7, pp. 830–832.

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". *Nat. Rev. Genet.* 14.9, pp. 661–673.

Philpott, Dana J, Matthew T Sorbara, Susan J Robertson, and Kenneth Croitoru (2014). "NOD proteins: regulators of inflammation in health and disease". *Nat. Rev. Immunol.* 14, pp. 9–23.

Picard, Capucine, Waleed Al-Herz, Aziz Bousfiha, Jean-Laurent Casanova, Talal Chatila, Mary Ellen Conley, Charlotte Cunningham-Rundles, Amos Etzioni, Steven M Holland, et al. (2015). "Primary Immunodeficiency Diseases: an Update on the Classification from the International Union of Immunological Societies Expert Committee for Primary Immunodeficiency 2015". *J. Clin. Immunol.* 35.8, pp. 696–726.

Pierre, Sandra, Thomas Eschenhagen, Gerd Geisslinger, and Klaus Scholich (2009). "Capturing adenylyl cyclases as potential drug targets". *Nat. Rev. Drug Discov.* 8.4, pp. 321–335.

Prescott, Natalie J, Benjamin Lehne, Kristina Stone, James C Lee, Kirstin Taylor, Jo Knight, Efterpi Papouli, Muddassar M Mirza, Michael A Simpson, et al. (2015). "Pooled sequencing of 531 genes in inflammatory bowel disease identifies an associated rare variant in BTNL2 and implicates other immune related genes". *PLoS Genet.* 11.2, e1004955.

Purcell, Shaun M, Jennifer L Moran, Menachem Fromer, Douglas Ruderfer, Nadia Solovieff, Panos Roussos, Colm O'Dushlaine, Kimberly Chambert, Sarah E Bergen, et al. (2014). "A polygenic burden of rare disruptive mutations in schizophrenia". *Nature* 506.7487, pp. 185–190.

Raj, Towfique, Katie Rothamel, Sara Mostafavi, Chun Ye, Mark N Lee, Joseph M Replogle, Ting Feng, Michelle Lee, Natasha Asinovski, et al. (2014). "Polarization of the effects of autoimmune and neurodegenerative risk alleles in leukocytes". *Science* 344.6183, pp. 519–523.

Raker, Verena Katharina, Christian Becker, and Kerstin Steinbrink (2016). "The cAMP Pathway as Therapeutic Target in Autoimmune and Inflammatory Diseases". *Front. Immunol.* 7, p. 123.

Rioux, John D, Ramnik J Xavier, Kent D Taylor, Mark S Silverberg, Philippe Goyette, Alan Huett, Todd Green, Petric Kuballa, M Michael Barmada, et al. (2007). "Genome-wide association study identifies new susceptibility loci for Crohn disease and implicates autophagy in disease pathogenesis". *Nat. Genet.* 39.5, pp. 596–604.

Risch, Neil and Kathleen Merikangas (1996). "The Future of Genetic Studies of Complex Human Diseases". *Science* 273, pp. 1516–1517.

Risøe, Petter K, Arkady Rutkovskiy, Joanna Ågren, Ingrid B M Kolseth, Signe Flood Kjeldsen, Guro Valen, Jarle Vaage, and Maria K Dahle (2015).

"Higher TNFα responses in young males compared to females are associated with attenuation of monocyte adenylyl cyclase expression". *Hum. Immunol.* 76.6, pp. 427–430.

Rivas, Manuel A, Mélissa Beaudoin, Agnes Gardet, Christine Stevens, Yashoda Sharma, Clarence K Zhang, Gabrielle Boucher, Stephan Ripke, David Ellinghaus, et al. (2011). "Deep resequencing of GWAS loci identifies independent rare variants associated with inflammatory bowel disease". *Nat. Genet.* 43.11, pp. 1066–1073.

Rivas, Manuel A, Daniel Graham, Patrick Sulem, Christine Stevens, A Nicole Desch, Philippe Goyette, Daniel Gudbjartsson, Ingileif Jonsdottir, Unnur Thorsteinsdottir, et al. (2016). "A protein-truncating R179X variant in RNF186 confers protection against ulcerative colitis". *Nat. Commun.* 7.

Roberts, R L, J E Hollis-Moffatt, R B Gearry, M A Kennedy, M L Barclay, and T R Merriman (2008). "Confirmation of association of IRGM and NCF4 with ileal Crohn's disease in a population-based cohort". *Genes Immun.* 9.6, pp. 561–565.

Rossi, Adriano G, Judith C Mc Cutcheon, Noemi Roy, Edwin R Chilvers, Christopher Haslett, and Ian Dransfield (1998). "Regulation of Macrophage Phagocytosis of Apoptotic Cells by cAMP1". *J. Immunol.* 160, pp. 3562–3568.

Rueda, Cesar M, Courtney M Jackson, and Claire A Chougnet (2016). "Regulatory T-Cell-Mediated Suppression of Conventional T-Cells and Dendritic Cells by Different cAMP Intracellular Pathways". *Front. Immunol.* 7, p. 216.

Sachidanandam, R, D Weissman, S C Schmidt, J M Kakol, L D Stein, G Marth, S Sherry, J C Mullikin, B J Mortimore, et al. (2001). "A map of human genome sequence variation containing 1.42 million single nucleotide polymorphisms". *Nature* 409.6822, pp. 928–933.

Sandborn, William J, Jean Frédéric Colombel, Roberts Enns, Brian G Feagan, Stephen B Hanauer, Ian C Lawrance, Remo Panaccione, Martin Sanders, Stefan Schreiber, et al. (2005). "Natalizumab induction and maintenance therapy for Crohn's disease". *N. Engl. J. Med.* 353.18, pp. 1912–1925.

Sandborn, William J, Christopher Gasink, Long-Long Gao, Marion A Blank, Jewel Johanns, Cynthia Guzzo, Bruce E Sands, Stephen B Hanauer, Stephan Targan, et al. (2012). "Ustekinumab induction and maintenance therapy in refractory Crohn's disease". *N. Engl. J. Med.* 367.16, pp. 1519–1528.

Sandborn, William J, Brian G Feagan, Paul Rutgeerts, Stephen Hanauer, Jean-Frédéric Colombel, Bruce E Sands, Milan Lukas, Richard N Fedorak, Scott Lee, et al. (2013). "Vedolizumab as induction and maintenance therapy for Crohn's disease". *N. Engl. J. Med.* 369.8, pp. 711–721.

Sands, Bruce E and Stacey Grabert (2009). "Epidemiology of Inflammatory Bowel Disease and Overview of Pathogenesis". *Med. Health R. I.* 92.3, pp. 73–77.

Sanger, F, S Nicklen, and A R Coulson (1977). "DNA sequencing with chain-terminating inhibitors". *Proc. Natl. Acad. Sci. U. S. A.* 74.12, pp. 5463–5467.

Satsangi, J, Ken I Welsh, Mike Bunce, Cecile Julier, J Mark Farrant, John I Bell, and Derek P Jewell (1996). "Contribution of genes of the major histocompatibility complex to susceptibility and disease phenotype in inflammatory bowel disease". *Lancet* 347, pp. 1212–1217.

Sawa, Shinichiro, Matthias Lochner, Naoko Satoh-Takayama, Sophie Dulauroy, Marion Bérard, Melanie Kleinschek, Daniel Cua, James P Di Santo, and Gérard Eberl (2011). "RORγt+ innate lymphoid cells regulate intestinal homeostasis by integrating negative signals from the symbiotic microbiota". *Nat. Immunol.* 12.4, pp. 320–326.

Schaefer, Carl F, Kira Anthony, Shiva Krupa, Jeffrey Buchoff, Matthew Day, Timo Hannay, and Kenneth H Buetow (2009). "PID: the Pathway Interaction Database". *Nucleic Acids Res.* 37.Database issue, pp. D674–9.

Schloss, Patrick D, Kathryn D Iverson, Joseph F Petrosino, and Sarah J Schloss (2014). "The dynamics of a family's gut microbiota reveal variations on a theme". *Microbiome* 2.1, p. 25.

Seed, Cotton, Alex Bloemendal, Jonathan M Bloom, Jacqueline I Goldstein, Daniel King, Timothy Poterba, and Benjamin M Neale (2017). *Hail: An Open-Source Framework for Scalable Genetic Data Analysis.* URL: `https://github.com/hail-is/hail`.

Seizinger, B R, G A Rouleau, L J Ozelius, A H Lane, A G Faryniarz, M V Chao, S Huson, B R Korf, D M Parry, et al. (1987). "Genetic linkage of von Recklinghausen neurofibromatosis to the nerve growth factor receptor gene". *Cell* 49.5, pp. 589–594.

Shah, T S, J Z Liu, J A B Floyd, J A Morris, N Wirth, J C Barrett, and C A Anderson (2012). "optiCall: a robust genotype-calling algorithm for rare, low-frequency and common variants". *Bioinformatics* 28.12, pp. 1598–1603.

Shih, David Q, Lola Y Kwan, Valerie Chavez, Offer Cohavy, Rivkah Gonsky, Elmer Y Chang, Christopher Chang, Charles O Elson, and Stephan R Targan (2009). "Microbial Induction of Inflammatory Bowel Disease Associated Gene TL1A (TNFSF15) in Antigen Presenting Cells". *Eur. J. Immunol.* 39.11, pp. 3239–3250.

Siddique, T, D A Figlewicz, M A Pericak-Vance, J L Haines, G Rouleau, A J Jeffers, P Sapp, W Y Hung, J Bebout, and D McKenna-Yasek (1991). "Linkage of a gene causing familial amyotrophic lateral sclerosis to chromosome 21 and evidence of genetic-locus heterogeneity". *N. Engl. J. Med.* 324.20, pp. 1381–1384.

Silverberg, Mark S, Lucia Mirea, Shelley B Bull, Janet E Murphy, A Hillary Steinhart, Gordon R Greenberg, Robin S McLeod, Zane Cohen, Judith A Wade, and Katherine A Siminovitch (2003). "A population- and family-based study of Canadian families reveals association of HLA DRB1*0103 with colonic involvement in inflammatory bowel disease". *Inflamm. Bowel Dis.* 9.1, pp. 1–9.

Silverberg, Mark S, Judy H Cho, John D Rioux, Dermot P B McGovern, Jing Wu, Vito Annese, Jean-Paul Achkar, Philippe Goyette, Regan Scott, et al. (2009). "Ulcerative colitis-risk loci on chromosomes 1p36 and 12q15 found by genome-wide association study". *Nat. Genet.* 41.2, pp. 216–220.

Sims, David, Ian Sudbery, Nicholas E Ilott, Andreas Heger, and Chris P Ponting (2014). "Sequencing depth and coverage: key considerations in genomic analyses". *Nat. Rev. Genet.* 15.2, pp. 121–132.

Skotte, Line, Thorfinn Sand Korneliussen, and Anders Albrechtsen (2012). "Association testing for next-generation sequencing data using score statistics". *Genet. Epidemiol.* 36.5, pp. 430–437.

Spain, Sarah L and Jeffrey C Barrett (2015). "Strategies for fine-mapping complex traits". *Hum. Mol. Genet.* 24.R1, R111–9.

Speer, M C, L H Yamaoka, J H Gilchrist, C P Gaskell, J M Stajich, J M Vance, A Kazantsev, A A Lastra, C S Haynes, and J S Beckmann (1992). "Confirmation of genetic heterogeneity in limb-girdle muscular dystrophy: linkage of an autosomal dominant form to chromosome 5q". *Am. J. Hum. Genet.* 50.6, pp. 1211–1217.

Stephens, Philip J, Patrick S Tarpey, Helen Davies, Peter Van Loo, Chris Greenman, David C Wedge, Serena Nik-Zainal, Sancha Martin, Ignacio Varela, et al. (2012). "The landscape of cancer genes and mutational processes in breast cancer". *Nature* 486.7403, pp. 400–404.

Sun, Jianping, Yingye Zheng, and Li Hsu (2013). "A unified mixed-effects model for rare-variant association in sequencing studies". *Genet. Epidemiol.* 37.4, pp. 334–344.

Suskind, David L, Mitchell J Brittnacher, Ghassan Wahbeh, Michele L Shaffer, Hillary S Hayden, Xuan Qin, Namita Singh, Christopher J Damman, Kyle R Hager, et al. (2015). "Fecal Microbial Transplant Effect on Clinical Outcomes and Fecal Microbiome in Active Crohns Disease". *Inflamm. Bowel Dis.* 21.3, pp. 556–563.

Tarazona-Santos, Eduardo, Moara Machado, Wagner C S Magalhães, Renee Chen, Fernanda Lyon, Laurie Burdett, Andrew Crenshaw, Cristina Fabbri, Latife Pereira, et al. (2013). "Evolutionary dynamics of the human NADPH oxidase genes CYBB, CYBA, NCF2, and NCF4: functional implications". *Mol. Biol. Evol.* 30.9, pp. 2157–2167.

The 1000 Genomes Project Consortium (2010). *1000 Genomes Project Phase I.* URL: ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/technical/reference/human_g1k_v37.fasta.gz.

– (2011). *1000 Genomes Project Phase II.* URL: ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/technical/reference/phase2_reference_assembly_sequence/hs37d5.fa.gz.

The International HapMap Consortium (2005). "A haplotype map of the human genome". *Nature* 437.7063, pp. 1299–1320.

The 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, pp. 661–678.

Tiemessen, Machteld M, Ann L Jagger, Hayley G Evans, Martijn J C van Herwijnen, Susan John, and Leonie S Taams (2007). "CD4+CD25+Foxp3+ regulatory T cells induce alternative activation of human monocytes/macrophages". *Proc. Natl. Acad. Sci. U. S. A.* 104.49, pp. 19446–19451.

Travis, Mark A, Boris Reizis, Andrew C Melton, Emma Masteller, Qizhi Tang, John M Proctor, Yanli Wang, Xin Bernstein, Xiaozhu Huang, et al. (2007). "Loss of integrin alpha(v)beta8 on dendritic cells causes autoimmunity and colitis in mice". *Nature* 449.7160, pp. 361–365.

Trynka, Gosia, Karen A Hunt, Nicholas A Bockett, Jihane Romanos, Vanisha Mistry, Agata Szperl, Sjoerd F Bakker, Maria Teresa Bardella, Leena Bhaw-rosun, et al. (2011). "Dense genotyping identifies and localizes multiple common and rare variant association signals in celiac disease". *Nat. Genet.* 43.12, pp. 1193–1201.

Tsui, Lap-Chee, Manuel Buchwald, David Barker, Jeffrey C Braman, Robert Knowlton, James W Schumm, Hans Eiberg, Jan Moher, Dara Kennedy, et al. (1985). "Cystic Fibrosis Locus Defined by a Genetically Linked Polymorphic Marker". *Science* 230, pp. 1054–1057.

Uhlig, Holm H (2013). "Monogenic diseases associated with intestinal inflammation: implications for the understanding of inflammatory bowel disease". *Gut* 62.12, pp. 1795–1805.

Van Limbergen, Johan, David C Wilson, and Jack Satsangi (2009). "The genetics of Crohn's disease". *Annu. Rev. Genomics Hum. Genet.* 10, pp. 89–116.

Vance, J M, G A Nicholson, L H Yamaoka, J Stajich, C S Stewart, M C Speer, W Y Hung, A D Roses, D Barker, and M A Pericak-Vance (1989). "Linkage of Charcot-Marie-Tooth neuropathy type 1a to chromosome 17". *Exp. Neurol.* 104.2, pp. 186–189.

Vermeire, Séverine, Sharon O'Byrne, Mary Keir, Marna Williams, Timothy T Lu, John C Mansfield, Christopher A Lamb, Brian G Feagan, Julian Panes, et al.

(2014). "Etrolizumab as induction therapy for ulcerative colitis: a randomised, controlled, phase 2 trial". *Lancet* 384.9940, pp. 309–318.

Visscher, Peter M, Sarah E Medland, Manuel A R Ferreira, Katherine I Morley, Gu Zhu, Belinda K Cornes, Grant W Montgomery, and Nicholas G Martin (2006). "Assumption-free estimation of heritability from genome-wide identity-by-descent sharing between full siblings". *PLoS Genet.* 2.3, e41.

Walter, Klaudia, Josine L Min, Jie Huang, Lucy Crooks, Yasin Memari, Shane McCarthy, John R B Perry, Changjiang Xu, Marta Futema, et al. (2015). "The UK10K project identifies rare variants in health and disease". *Nature* 526.7571, pp. 82–90.

Wang, Guoxing, Ana C Abadía-Molina, Scott B Berger, Xavier Romero, Michael S O'Keeffe, Domingo I Rojas-Barros, Marta Aleman, Gongxian Liao, Elena Maganto-García, et al. (2012). "Cutting edge: Slamf8 is a negative regulator of Nox2 activity in macrophages". *J. Immunol.* 188.12, pp. 5829–5832.

Wang, Guoxing, Boaz J van Driel, Gongxian Liao, Michael S O'Keeffe, Peter J Halibozek, Jacky Flipse, Burcu Yigit, Veronica Azcutia, Francis W Luscinskas, et al. (2015). "Migration of myeloid cells during inflammation is differentially regulated by the cell surface receptors Slamf1 and Slamf8". *PLoS One* 10.3, e0121968.

Weaver, Casey T and Robin D Hatton (2009). "Interplay between the TH17 and TReg cell lineages: a (co-)evolutionary perspective". *Nat. Rev. Immunol.* 9.12, pp. 883–889.

Weber, J L and P E May (1989). "Abundant class of human DNA polymorphisms which can be typed using the polymerase chain reaction". *Am. J. Hum. Genet.* 44.3, pp. 388–396.

Wellcome Trust Case Control Consortium, Julian B Maller, Gilean McVean, Jake Byrnes, Damjan Vukcevic, Kimmo Palin, Zhan Su, Joanna M M Howson, Adam Auton, et al. (2012). "Bayesian refinement of association signals for 14 loci in 3 common diseases". *Nat. Genet.* 44.12, pp. 1294–1301.

Westra, Harm-Jan, Marjolein J Peters, Tõnu Esko, Hanieh Yaghootkar, Claudia Schurmann, Johannes Kettunen, Mark W Christiansen, Benjamin P Fairfax, Katharina Schramm, et al. (2013). "Systematic identification of trans eQTLs as putative drivers of known disease associations". *Nat. Genet.* 45.10, pp. 1238–1243.

Willer, Cristen J, Yun Li, and Gonçalo R Abecasis (2010). "METAL: fast and efficient meta-analysis of genomewide association scans". *Bioinformatics* 26.17, pp. 2190–2191.

Withers, David R, Matthew R Hepworth, Xinxin Wang, Emma C Mackley, Emily E Halford, Emma E Dutton, Clare L Marriott, Verena Brucklacher-Waldert, Marc Veldhoen, et al. (2016). "Transient inhibition

of ROR-[gamma]t therapeutically limits intestinal inflammation by reducing TH17 cells and preserving group 3 innate lymphoid cells". *Nat. Med.* 22.3, pp. 319–323.

Worthington, John J, Aoife Kelly, Catherine Smedley, David Bauché, Simon Campbell, Julien C Marie, and Mark A Travis (2015). "Integrin αvβ8-Mediated TGF-β Activation by Effector Regulatory T Cells Is Essential for Suppression of T-Cell-Mediated Inflammation". *Immunity* 42.5, pp. 903–915.

Wright, Caroline F, Tomas W Fitzgerald, Wendy D Jones, Stephen Clayton, Jeremy F McRae, Margriet van Kogelenberg, Daniel A King, Kirsty Ambridge, Daniel M Barrett, et al. (2015). "Genetic diagnosis of developmental disorders in the DDD study: a scalable analysis of genome-wide research data". *Lancet* 385.9975, pp. 1305–1314.

Wright, Fred A, Patrick F Sullivan, Andrew I Brooks, Fei Zou, Wei Sun, Kai Xia, Vered Madar, Rick Jansen, Wonil Chung, et al. (2014). "Heritability and genomics of gene expression in peripheral blood". *Nat. Genet.* 46.5, pp. 430–437.

Wu, Michael C, Seunggeun Lee, Tianxi Cai, Yun Li, Michael Boehnke, and Xihong Lin (2011). "Rare-variant association testing for sequencing data with the sequence kernel association test". *Am. J. Hum. Genet.* 89.1, pp. 82–93.

Yamazaki, Keiko, Masakazu Takazoe, Torao Tanaka, Toshiki Kazumori, and Yusuke Nakamura (2002). "Absence of mutation in the NOD2 / CARD15 gene among 483 Japanese patients with Crohns disease". *J. Hum. Genet.* 47, pp. 469–472.

Yamazaki, Keiko, Masakazu Takazoe, Torao Tanaka, Toshiki Ichimori, Susumu Saito, Aritoshi Iida, Yoshihiro Onouchi, Akira Hata, and Yusuke Nakamura (2004). "Association analysis of SLC22A4 , SLC22A5 and DLG5 in Japanese patients with Crohn disease". *J. Hum. Genet.* 49, pp. 664–668.

Yamazaki, Keiko, Dermot McGovern, Jiannis Ragoussis, Marta Paolucci, Helen Butler, Derek Jewell, Lon Cardon, Masakazu Takazoe, Torao Tanaka, et al. (2005). "Single nucleotide polymorphisms in TNFSF15 confer susceptibility to Crohn's disease". *Hum. Mol. Genet.* 14.22, pp. 3499–3506.

Yamazaki, Keiko, Junji Umeno, Atsushi Takahashi, Atsushi Hirano, Todd Andrew Johnson, Natsuhiko Kumasaka, Takashi Morizono, Naoya Hosono, Takaaki Kawaguchi, et al. (2013). "A genome-wide association study identifies 2 susceptibility Loci for Crohn's disease in a Japanese population". *Gastroenterology* 144.4, pp. 781–788.

Yang, Suk-Kyun, Myunghee Hong, Wanting Zhao, Yusun Jung, Naeimeh Tayebi, Byong Duk Ye, Kyung-Jo Kim, Sang Hyoung Park, Inchul Lee, et al. (2013). "Genome-Wide Association Study of Ulcerative Colitis in Koreans Suggests

Extensive Overlapping of Genetic Susceptibility With Caucasians". *Inflamm. Bowel Dis.* 19.5, pp. 954–966.

Yang, Suk-Kyun, Myunghee Hong, Jiwon Baek, Hyunchul Choi, Wanting Zhao, Yusun Jung, Talin Haritunians, Byong Duk Ye, Kyung-Jo Kim, et al. (2014a). "A common missense variant in NUDT15 confers susceptibility to thiopurine-induced leukopenia". *Nat. Genet.* 46.9, pp. 1017–1020.

Yang, Suk-Kyun, Myunghee Hong, Wanting Zhao, Yusun Jung, Jiwon Baek, Naeimeh Tayebi, Kyung Mo Kim, Byong Duk Ye, Kyung-Jo Kim, et al. (2014b). "Genome-wide association study of Crohn's disease in Koreans revealed three new susceptibility loci and common attributes of genetic susceptibility across ethnic populations". *Gut* 63.1, pp. 80–87.

Yang, Yi, Miriam B Torchinsky, Michael Gobert, Huizhong Xiong, Mo Xu, Jonathan L Linehan, Francis Alonzo, Charles Ng, Alessandra Chen, et al. (2014c). "Focused specificity of intestinal TH17 cells towards commensal bacterial antigens". *Nature* 510.7503, pp. 152–156.

Ye, Chun Jimmie, Ting Feng, Ho-Keun Kwon, Towfique Raj, Michael T Wilson, Natasha Asinovski, Cristin McCabe, Michelle H Lee, Irene Frohlich, et al. (2014). "Intersection of population variation and autoimmunity genetics in human T cell activation". *Science* 345.6202, p. 1254665.

Zeissig, Yvonne, Britt-Sabina Petersen, Snezana Milutinovic, Esther Bosse, Gabriele Mayr, Kenneth Peuker, Jelka Hartwig, Andreas Keller, Martina Kohl, et al. (2015). "XIAP variants in male Crohn's disease". *Gut* 64.1, pp. 66–76.

Zeller, Tanja, Philipp Wild, Silke Szymczak, Maxime Rotival, Arne Schillert, Raphaele Castagne, Seraya Maouche, Marine Germain, Karl Lackner, et al. (2010). "Genetics and Beyond – The Transcriptome of Human Monocytes and Disease Susceptibility". *PLoS One* 5.5, e10693.

Zhernakova, Alexandra, Cleo C van Diemen, and Cisca Wijmenga (2009). "Detecting shared pathogenesis from the shared genetics of immune-related diseases". *Nat. Rev. Genet.* 10.1, pp. 43–55.

Zhernakova, Daria, Patrick Deelen, Martijn Vermaat, Maarten van Iterson, Michiel van Galen, Wibowo Arindrarto, Peter van t Hof, Hailiang Mei, Freerk van Dijk, et al. (2015). "Hypothesis-free identification of modulators of genetic risk factors".

Zhou, Qing, Geun-Shik Lee, Jillian Brady, Shrimati Datta, Matilda Katan, Afzal Sheikh, Marta S Martins, Tom D Bunney, Brian H Santich, et al. (2012). "A hypermorphic missense mutation in PLCG2, encoding phospholipase Cγ2, causes a dominantly inherited autoinflammatory disease with immunodeficiency". *Am. J. Hum. Genet.* 91.4, pp. 713–720.

Zimmerman, Noah P, Suresh N Kumar, Jerrold R Turner, and Michael B Dwinell (2012). "Cyclic AMP dysregulates intestinal epithelial cell restitution through PKA and RhoA". *Inflamm. Bowel Dis.* 18.6, pp. 1081–1091.

Zuk, Or, Stephen F Schaffner, Kaitlin Samocha, Ron Do, Eliana Hechter, Sekar Kathiresan, Mark J Daly, Benjamin M Neale, Shamil R Sunyaev, and Eric S Lander (2014). "Searching for missing heritability: designing rare variant association studies". *Proc. Natl. Acad. Sci. U. S. A.* 111, E455–64.