

References

- 1 . Abecasis, G. R., A. Auton, L. D. Brooks, M. A. DePristo, R. M. Durbin, R. E. Handsaker, . . . G. A. McVean (2012). "An integrated map of genetic variation from 1,092 human genomes." Nature **491**(7422): 56-65.
- 2 . Abifadel, M., J. P. Rabes, M. Devillers, A. Munnich, D. Erlich, C. Junien, . . . C. Boileau (2009). "Mutations and polymorphisms in the proprotein convertase subtilisin kexin 9 (PCSK9) gene in cholesterol metabolism and disease." Hum Mutat **30**(4): 520-529.
- 3 . Abifadel, M., M. Varret, J. P. Rabes, D. Allard, K. Ouguerram, M. Devillers, . . . C. Boileau (2003). "Mutations in PCSK9 cause autosomal dominant hypercholesterolemia." Nat Genet **34**(2): 154-156.
- 4 . Akinsheye, I., A. Alsultan, N. Solovieff, D. Ngo, C. T. Baldwin, P. Sebastiani, . . . M. H. Steinberg (2011). "Fetal hemoglobin in sickle cell anemia." Blood **118**(1): 19-27.
- 5 . Albert, T. J., M. N. Molla, D. M. Muzny, L. Nazareth, D. Wheeler, X. Song, . . . R. A. Gibbs (2007). "Direct selection of human genomic loci by microarray hybridization." Nat Methods **4**(11): 903-905.
- 6 . Allard, D., S. Amsellem, M. Abifadel, M. Trillard, M. Devillers, G. Luc, . . . J. P. Rabes (2005). "Novel mutations of the PCSK9 gene cause variable phenotype of autosomal dominant hypercholesterolemia." Hum Mutat **26**(5): 497.
- 7 . Amarenco, P., P. Lavallee and P. J. Touboul (2004). "Stroke prevention, blood cholesterol, and statins." Lancet Neurol **3**(5): 271-278.
- 8 . Amarenco, P. and P. G. Steg (2007). "The paradox of cholesterol and stroke." Lancet **370**(9602): 1803-1804.
- 9 . Anderson, G. L., J. Manson, R. Wallace, B. Lund, D. Hall, S. Davis, . . . R. L. Prentice (2003). "Implementation of the Women's Health Initiative study design." Ann Epidemiol **13**(9 Suppl): S5-17.
- 10 . Anderson, K. M., W. P. Castelli and D. Levy (1987). "Cholesterol and mortality. 30 years of follow-up from the Framingham study." JAMA **257**(16): 2176-2180.
- 11 . Arsenault, B. J., S. M. Boekholdt and J. J. Kastelein (2011). "Lipid parameters for measuring risk of cardiovascular disease." Nat Rev Cardiol **8**(4): 197-206.
- 12 . Arsenault, B. J., J. S. Rana, E. S. Stroes, J. P. Despres, P. K. Shah, J. J. Kastelein, . . . K. T. Khaw (2009). "Beyond low-density lipoprotein cholesterol: respective contributions of non-high-density lipoprotein cholesterol levels, triglycerides, and the total cholesterol/high-density lipoprotein cholesterol ratio to coronary heart disease risk in apparently healthy men and women." J Am Coll Cardiol **55**(1): 35-41.
- 13 . Asimit, J. and E. Zeggini (2010). "Rare variant association analysis methods for complex traits." Annu Rev Genet **44**: 293-308.
- 14 . Aslibekyan, S., E. K. Kabagambe, M. R. Irvin, R. J. Straka, I. B. Borecki, H. K. Tiwari, . . . D. K. Arnett (2012). "A genome-wide association study of inflammatory biomarker changes in response to fenofibrate treatment in the Genetics of Lipid Lowering Drug and Diet Network." Pharmacogenet Genomics **22**(3): 191-197.
- 15 . Assmann, G., H. Schulte, A. von Eckardstein and Y. Huang (1996). "High-density lipoprotein cholesterol as a predictor of coronary heart disease risk. The PROCAM experience and pathophysiological implications for reverse cholesterol transport." Atherosclerosis **124** Suppl: S11-20.
- 16 . Aulchenko, Y. S., S. Ripatti, I. Lindqvist, D. Boomsma, I. M. Heid, P. P. Pramstaller, . . . E. Consortium (2009). "Loci influencing lipid levels and coronary heart disease risk in 16 European population cohorts." Nat Genet **41**(1): 47-55.
- 17 . Badimon, J. J., C. G. Santos-Gallego and L. Badimon (2010). "[Importance of HDL cholesterol in atherothrombosis: how did we get here? Where are we going?]." Rev Esp Cardiol **63** Suppl 2: 20-35.
- 18 . Badimon, L. and G. Vilahur (2012). "LDL-cholesterol versus HDL-cholesterol in the atherosclerotic plaque: inflammatory resolution versus thrombotic chaos." Ann N Y Acad Sci **1254**: 18-32.
- 19 . Baigent, C., A. Keech, P. M. Kearney, L. Blackwell, G. Buck, C. Pollicino, . . . C. Cholesterol Treatment Trialists (2005). "Efficacy and safety of cholesterol-lowering treatment: prospective meta-

analysis of data from 90,056 participants in 14 randomised trials of statins." *Lancet* **366**(9493): 1267-1278.

20 . Bak, S., D. Gaist, S. H. Sindrup, A. Skytthe and K. Christensen (2002). "Genetic liability in stroke: a long-term follow-up study of Danish twins." *Stroke* **33**(3): 769-774.

21 . Ballester, B., A. Medina-Rivera, D. Schmidt, M. Gonzalez-Porta, M. Carlucci, X. Chen, . . . M. D. Wilson (2014). "Multi-species, multi-transcription factor binding highlights conserved control of tissue-specific biological pathways." *Elife* **3**: e02626.

22 . Bamshad, M. J., S. B. Ng, A. W. Bigham, H. K. Tabor, M. J. Emond, D. A. Nickerson and J. Shendure (2011). "Exome sequencing as a tool for Mendelian disease gene discovery." *Nat Rev Genet* **12**(11): 745-755.

23 . Bansal, S., J. E. Buring, N. Rifai, S. Mora, F. M. Sacks and P. M. Ridker (2007). "Fasting compared with nonfasting triglycerides and risk of cardiovascular events in women." *JAMA* **298**(3): 309-316.

24 . Barreiro, L. B., L. Tailleux, A. A. Pai, B. Gicquel, J. C. Marioni and Y. Gilad (2012). "Deciphering the genetic architecture of variation in the immune response to Mycobacterium tuberculosis infection." *Proc Natl Acad Sci U S A* **109**(4): 1204-1209.

25 . Barrett, J. C. and L. R. Cardon (2006). "Evaluating coverage of genome-wide association studies." *Nat Genet* **38**(6): 659-662.

26 . Barter, P. (2009). "Lessons learned from the Investigation of Lipid Level Management to Understand its Impact in Atherosclerotic Events (ILLUMINATE) trial." *Am J Cardiol* **104**(10 Suppl): 10E-15E.

27 . Barter, P. J., M. Caulfield, M. Eriksson, S. M. Grundy, J. J. Kastelein, M. Komajda, . . . I. Investigators (2007). "Effects of torcetrapib in patients at high risk for coronary events." *N Engl J Med* **357**(21): 2109-2122.

28 . Barton, A., W. Thomson, X. Ke, S. Eyre, A. Hinks, J. Bowes, . . . J. Worthington (2008). "Rheumatoid arthritis susceptibility loci at chromosomes 10p15, 12q13 and 22q13." *Nat Genet* **40**(10): 1156-1159.

29 . Basel-Vanagaite, L., N. Zevit, A. Har Zahav, L. Guo, S. Parathath, M. Pasmanik-Chor, . . . R. Shamir (2012). "Transient infantile hypertriglyceridemia, fatty liver, and hepatic fibrosis caused by mutated GPD1, encoding glycerol-3-phosphate dehydrogenase 1." *Am J Hum Genet* **90**(1): 49-60.

30 . Bauer, D. E. and S. H. Orkin (2011). "Update on fetal hemoglobin gene regulation in hemoglobinopathies." *Curr Opin Pediatr* **23**(1): 1-8.

31 . Beekman, M., B. T. Heijmans, N. G. Martin, N. L. Pedersen, J. B. Whitfield, U. DeFaire, . . . D. I. Boomsma (2002). "Heritabilities of apolipoprotein and lipid levels in three countries." *Twin Res* **5**(2): 87-97.

32 . Beigneux, A. P., R. Franssen, A. Bensadoun, P. Gin, K. Melford, J. Peter, . . . S. G. Young (2009). "Chylomicronemia with a mutant GPIHBP1 (Q115P) that cannot bind lipoprotein lipase." *Arterioscler Thromb Vasc Biol* **29**(6): 956-962.

33 . Bell, G. I., S. Horita and J. H. Karam (1984). "A polymorphic locus near the human insulin gene is associated with insulin-dependent diabetes mellitus." *Diabetes* **33**(2): 176-183.

34 . Benyamin, B., M. A. Ferreira, G. Willemsen, S. Gordon, R. P. Middelberg, B. P. McEvoy, . . . J. B. Whitfield (2009). "Common variants in Tmprss6 are associated with iron status and erythrocyte volume." *Nat Genet* **41**(11): 1173-1175.

35 . Benyamin, B., A. F. McRae, G. Zhu, S. Gordon, A. K. Henders, A. Palotie, . . . P. M. Visscher (2009). "Variants in TF and HFE explain approximately 40% of genetic variation in serum-transferrin levels." *Am J Hum Genet* **84**(1): 60-65.

36 . Berge, K. E., H. Tian, G. A. Graf, L. Yu, N. V. Grishin, J. Schultz, . . . H. H. Hobbs (2000). "Accumulation of dietary cholesterol in sitosterolemia caused by mutations in adjacent ABC transporters." *Science* **290**(5497): 1771-1775.

37 . Biomarkers Definitions Working, G. (2001). "Biomarkers and surrogate endpoints: preferred definitions and conceptual framework." *Clin Pharmacol Ther* **69**(3): 89-95.

- 38** . Blair, D. R., C. S. Lyttle, J. M. Mortensen, C. F. Bearden, A. B. Jensen, H. Khiabani, . . . A. Rzhetsky (2013). "A nondegenerate code of deleterious variants in Mendelian loci contributes to complex disease risk." *Cell* **155**(1): 70-80.
- 39** . Boekholdt, S. M. and J. J. Kastelein (2010). "C-reactive protein and cardiovascular risk: more fuel to the fire." *Lancet* **375**(9709): 95-96.
- 40** . Bonnelykke, K., M. C. Matheson, T. H. Pers, R. Granell, D. P. Strachan, A. C. Alves, . . . C. Lifecourse Epidemiology (2013). "Meta-analysis of genome-wide association studies identifies ten loci influencing allergic sensitization." *Nat Genet* **45**(8): 902-906.
- 41** . Bostom, A. G., L. A. Cupples, J. L. Jenner, J. M. Ordovas, L. J. Seman, P. W. Wilson, . . . W. P. Castelli (1996). "Elevated plasma lipoprotein(a) and coronary heart disease in men aged 55 years and younger. A prospective study." *JAMA* **276**(7): 544-548.
- 42** . Botstein, D. and N. Risch (2003). "Discovering genotypes underlying human phenotypes: past successes for mendelian disease, future approaches for complex disease." *Nat Genet* **33** **Suppl**: 228-237.
- 43** . Boycott, K. M., M. R. Vanstone, D. E. Bulman and A. E. MacKenzie (2013). "Rare-disease genetics in the era of next-generation sequencing: discovery to translation." *Nat Rev Genet* **14**(10): 681-691.
- 44** . British Cardiac, S., S. British Hypertension, U. K. Diabetes, U. K. Heart, S. Primary Care Cardiovascular and A. Stroke (2005). "JBS 2: Joint British Societies' guidelines on prevention of cardiovascular disease in clinical practice." *Heart* **91** **Suppl 5**: v1-52.
- 45** . Brotman, D. J., E. Walker, M. S. Lauer and R. G. O'Brien (2005). "In search of fewer independent risk factors." *Arch Intern Med* **165**(2): 138-145.
- 46** . Brown, M. L., A. Inazu, C. B. Hesler, L. B. Agellon, C. Mann, M. E. Whitlock, . . . et al. (1989). "Molecular basis of lipid transfer protein deficiency in a family with increased high-density lipoproteins." *Nature* **342**(6248): 448-451.
- 47** . Brown, M. S. and J. L. Goldstein (1976). "Receptor-mediated control of cholesterol metabolism." *Science* **191**(4223): 150-154.
- 48** . Browning, B. L. and S. R. Browning (2009). "A unified approach to genotype imputation and haplotype-phase inference for large data sets of trios and unrelated individuals." *Am J Hum Genet* **84**(2): 210-223.
- 49** . Brunner, E. J., M. Kivimaki, D. R. Witte, D. A. Lawlor, G. Davey Smith, J. A. Cooper, . . . M. Kumari (2008). "Inflammation, insulin resistance, and diabetes--Mendelian randomization using CRP haplotypes points upstream." *PLoS Med* **5**(8): e155.
- 50** . Buijssse, B., R. K. Simmons, S. J. Griffin and M. B. Schulze (2011). "Risk assessment tools for identifying individuals at risk of developing type 2 diabetes." *Epidemiol Rev* **33**(1): 46-62.
- 51** . Burkhardt, R., E. E. Kenny, J. K. Lowe, A. Birkeland, R. Josowitz, M. Noel, . . . J. L. Breslow (2008). "Common SNPs in HMGCR in micronesians and whites associated with LDL-cholesterol levels affect alternative splicing of exon13." *Arterioscler Thromb Vasc Biol* **28**(11): 2078-2084.
- 52** . Campbell, P. J., C. MacLean, P. A. Beer, G. Buck, K. Wheatley, J. J. Kiladjian, . . . A. R. Green (2012). "Correlation of blood counts with vascular complications in essential thrombocythemia: analysis of the prospective PT1 cohort." *Blood* **120**(7): 1409-1411.
- 53** . CARDIoGRAMplusC4D Consortium (2015). "A Comprehensive 1000 Genomes-based GWAS meta-analysis of Coronary Artery Disease." under review.
- 54** . Cartier, A., M. Cote, I. Lemieux, L. Perusse, A. Tremblay, C. Bouchard and J. P. Despres (2009). "Age-related differences in inflammatory markers in men: contribution of visceral adiposity." *Metabolism* **58**(10): 1452-1458.
- 55** . Casas, J. P., T. Shah, J. Cooper, E. Hawe, A. D. McMahon, D. Gaffney, . . . A. D. Hingorani (2006). "Insight into the nature of the CRP-coronary event association using Mendelian randomization." *Int J Epidemiol* **35**(4): 922-931.
- 56** . Castelli, W. P. (1988). "Cholesterol and lipids in the risk of coronary artery disease--the Framingham Heart Study." *Can J Cardiol* **4** **Suppl A**: 5A-10A.

- 57** . Chambers, J. C., W. Zhang, Y. Li, J. Sehmi, M. N. Wass, D. Zabaneh, . . . J. S. Kooner (2009). "Genome-wide association study identifies variants in TM6RS6 associated with hemoglobin levels." *Nat Genet* **41**(11): 1170-1172.
- 58** . Chapman, J. M., J. D. Cooper, J. A. Todd and D. G. Clayton (2003). "Detecting disease associations due to linkage disequilibrium using haplotype tags: a class of tests and the determinants of statistical power." *Hum Hered* **56**(1-3): 18-31.
- 59** . Chasman, D. I., G. Pare, S. Mora, J. C. Hopewell, G. Peloso, R. Clarke, . . . P. M. Ridker (2009). "Forty-three loci associated with plasma lipoprotein size, concentration, and cholesterol content in genome-wide analysis." *PLoS Genet* **5**(11): e1000730.
- 60** . Chasman, D. I., G. Pare, R. Y. Zee, A. N. Parker, N. R. Cook, J. E. Buring, . . . P. M. Ridker (2008). "Genetic loci associated with plasma concentration of low-density lipoprotein cholesterol, high-density lipoprotein cholesterol, triglycerides, apolipoprotein A1, and Apolipoprotein B among 6382 white women in genome-wide analysis with replication." *Circ Cardiovasc Genet* **1**(1): 21-30.
- 61** . Chen, Z., H. Tang, R. Qayyum, U. M. Schick, M. A. Nalls, R. Handsaker, . . . A. P. Reiner (2013). "Genome-wide association analysis of red blood cell traits in African Americans: the COGENT Network." *Hum Mol Genet* **22**(12): 2529-2538.
- 62** . Cheng, T. L., Y. T. Wu, H. Y. Lin, F. C. Hsu, S. K. Liu, B. I. Chang, . . . H. L. Wu (2011). "Functions of rhomboid family protease RHBDL2 and thrombomodulin in wound healing." *J Invest Dermatol* **131**(12): 2486-2494.
- 63** . Choi, B. G., G. Vilahur, J. F. Viles-Gonzalez and J. J. Badimon (2006). "The role of high-density lipoprotein cholesterol in atherothrombosis." *Mt Sinai J Med* **73**(4): 690-701.
- 64** . Cholesterol Treatment Trialists, C., C. Baigent, L. Blackwell, J. Emberson, L. E. Holland, C. Reith, . . . R. Collins (2010). "Efficacy and safety of more intensive lowering of LDL cholesterol: a meta-analysis of data from 170,000 participants in 26 randomised trials." *Lancet* **376**(9753): 1670-1681.
- 65** . Cirulli, E. T. and D. B. Goldstein (2010). "Uncovering the roles of rare variants in common disease through whole-genome sequencing." *Nat Rev Genet* **11**(6): 415-425.
- 66** . Cladaras, C., M. Hadzopoulou-Cladaras, B. K. Felber, G. Pavlakis and V. I. Zannis (1987). "The molecular basis of a familial apoE deficiency. An acceptor splice site mutation in the third intron of the deficient apoE gene." *J Biol Chem* **262**(5): 2310-2315.
- 67** . Clarke, R., J. F. Peden, J. C. Hopewell, T. Kyriakou, A. Goel, S. C. Heath, . . . M. Farrall (2009). "Genetic variants associated with Lp(a) lipoprotein level and coronary disease." *N Engl J Med* **361**(26): 2518-2528.
- 68** . Coelho, H. C., S. C. Lopes, J. P. Pimentel, P. A. Nogueira, F. T. Costa, A. M. Siqueira, . . . M. V. Lacerda (2013). "Thrombocytopenia in Plasmodium vivax malaria is related to platelets phagocytosis." *PLoS One* **8**(5): e63410.
- 69** . Cohen, J., A. Pertsemlidis, I. K. Kotowski, R. Graham, C. K. Garcia and H. H. Hobbs (2005). "Low LDL cholesterol in individuals of African descent resulting from frequent nonsense mutations in PCSK9." *Nat Genet* **37**(2): 161-165.
- 70** . Cohen, J. C., E. Boerwinkle, T. H. Mosley, Jr. and H. H. Hobbs (2006). "Sequence variations in PCSK9, low LDL, and protection against coronary heart disease." *N Engl J Med* **354**(12): 1264-1272.
- 71** . Cohen, J. C., R. S. Kiss, A. Pertsemlidis, Y. L. Marcel, R. McPherson and H. H. Hobbs (2004). "Multiple rare alleles contribute to low plasma levels of HDL cholesterol." *Science* **305**(5685): 869-872.
- 72** . Companiononi, O., F. Rodriguez Esparragon, A. M. Fernandez-Aceituno and J. C. Rodriguez Perez (2011). "[Genetic variants, cardiovascular risk and genome-wide association studies]." *Rev Esp Cardiol* **64**(6): 509-514.
- 73** . Consortium, C. A. D., P. Deloukas, S. Kanoni, C. Willenborg, M. Farrall, T. L. Assimes, . . . N. J. Samani (2013). "Large-scale association analysis identifies new risk loci for coronary artery disease." *Nat Genet* **45**(1): 25-33.

- 74** . Coram, M. A., Q. Duan, T. J. Hoffmann, T. Thornton, J. W. Knowles, N. A. Johnson, . . . H. Tang (2013). "Genome-wide characterization of shared and distinct genetic components that influence blood lipid levels in ethnically diverse human populations." *Am J Hum Genet* **92**(6): 904-916.
- 75** . Coronary Artery Disease Genetics, C. (2011). "A genome-wide association study in Europeans and South Asians identifies five new loci for coronary artery disease." *Nat Genet* **43**(4): 339-344.
- 76** . Coviello, A. D., R. Haring, M. Wellons, D. Vaidya, T. Lehtimaki, S. Keildson, . . . J. R. Perry (2012). "A genome-wide association meta-analysis of circulating sex hormone-binding globulin reveals multiple Loci implicated in sex steroid hormone regulation." *PLoS Genet* **8**(7): e1002805.
- 77** . Cox, D. W., W. C. Breckenridge and J. A. Little (1978). "Inheritance of apolipoprotein C-II deficiency with hypertriglyceridemia and pancreatitis." *N Engl J Med* **299**(26): 1421-1424.
- 78** . Crosslin, D. R., A. McDavid, N. Weston, S. C. Nelson, X. Zheng, E. Hart, . . . N. Genomics (2012). "Genetic variants associated with the white blood cell count in 13,923 subjects in the eMERGE Network." *Hum Genet* **131**(4): 639-652.
- 79** . Crosslin, D. R., A. McDavid, N. Weston, X. Zheng, E. Hart, M. de Andrade, . . . N. Genomics (2013). "Genetic variation associated with circulating monocyte count in the eMERGE Network." *Hum Mol Genet* **22**(10): 2119-2127.
- 80** . Cushman, M., A. M. Arnold, B. M. Psaty, T. A. Manolio, L. H. Kuller, G. L. Burke, . . . R. P. Tracy (2005). "C-reactive protein and the 10-year incidence of coronary heart disease in older men and women: the cardiovascular health study." *Circulation* **112**(1): 25-31.
- 81** . D'Agostino, R. B., Sr., R. S. Vasan, M. J. Pencina, P. A. Wolf, M. Cobain, J. M. Massaro and W. B. Kannel (2008). "General cardiovascular risk profile for use in primary care: the Framingham Heart Study." *Circulation* **117**(6): 743-753.
- 82** . Danecek, P., A. Auton, G. Abecasis, C. A. Albers, E. Banks, M. A. DePristo, . . . G. Genomes Project Analysis (2011). "The variant call format and VCFtools." *Bioinformatics* **27**(15): 2156-2158.
- 83** . Danesh, J., R. Collins, P. Appleby and R. Peto (1998). "Association of fibrinogen, C-reactive protein, albumin, or leukocyte count with coronary heart disease: meta-analyses of prospective studies." *JAMA* **279**(18): 1477-1482.
- 84** . Danesh, J., J. G. Wheeler, G. M. Hirschfield, S. Eda, G. Eiriksdottir, A. Rumley, . . . V. Gudnason (2004). "C-reactive protein and other circulating markers of inflammation in the prediction of coronary heart disease." *N Engl J Med* **350**(14): 1387-1397.
- 85** . Dawber, T. R., W. B. Kannel, N. Revotskie, J. Stokes, 3rd, A. Kagan and T. Gordon (1959). "Some factors associated with the development of coronary heart disease: six years' follow-up experience in the Framingham study." *Am J Public Health Nations Health* **49**: 1349-1356.
- 86** . Degoma, E. M. and D. J. Rader (2011). "Novel HDL-directed pharmacotherapeutic strategies." *Nat Rev Cardiol* **8**(5): 266-277.
- 87** . Dehghan, A., J. Dupuis, M. Barbalic, J. C. Bis, G. Eiriksdottir, C. Lu, . . . D. I. Chasman (2011). "Meta-analysis of genome-wide association studies in >80 000 subjects identifies multiple loci for C-reactive protein levels." *Circulation* **123**(7): 731-738.
- 88** . Delaneau, O., J. F. Zagury and J. Marchini (2013). "Improved whole-chromosome phasing for disease and population genetic studies." *Nat Methods* **10**(1): 5-6.
- 89** . Dendrou, C. A., V. Plagnol, E. Fung, J. H. Yang, K. Downes, J. D. Cooper, . . . L. S. Wicker (2009). "Cell-specific protein phenotypes for the autoimmune locus IL2RA using a genotype-selectable human bioresource." *Nat Genet* **41**(9): 1011-1015.
- 90** . Ding, K., M. de Andrade, T. A. Manolio, D. C. Crawford, L. J. Rasmussen-Torvik, M. D. Ritchie, . . . I. J. Kullo (2013). "Genetic variants that confer resistance to malaria are associated with red blood cell traits in African-Americans: an electronic medical record-based genome-wide association study." *G3 (Bethesda)* **3**(7): 1061-1068.
- 91** . Do, R., N. O. Stitzel, H. Won, A. B. Jorgensen, S. Duga, P. Angelica Merlini, . . . S. Kathiresan (2014). "Exome sequencing identifies rare LDLR and APOA5 alleles conferring risk for myocardial infarction." *Nature*.

- 92** . Do, R., N. O. Stitzel, H. H. Won, A. B. Jorgensen, S. Duga, P. Angelica Merlini, . . . S. Kathiresan (2015). "Exome sequencing identifies rare LDLR and APOA5 alleles conferring risk for myocardial infarction." *Nature* **518**(7537): 102-106.
- 93** . Do, R., C. J. Willer, E. M. Schmidt, S. Sengupta, C. Gao, G. M. Peloso, . . . S. Kathiresan (2013). "Common variants associated with plasma triglycerides and risk for coronary artery disease." *Nat Genet* **45**(11): 1345-1352.
- 94** . Dorajoo, R., R. Li, M. K. Ikram, J. Liu, P. Froguel, J. Lee, . . . Y. Friedlander (2013). "Are C-reactive protein associated genetic variants associated with serum levels and retinal markers of microvascular pathology in Asian populations from Singapore?" *PLoS One* **8**(7): e67650.
- 95** . Doumatey, A. P., G. Chen, F. Tekola Ayele, J. Zhou, M. Erdos, D. Shriner, . . . C. N. Rotimi (2012). "C-reactive protein (CRP) promoter polymorphisms influence circulating CRP levels in a genome-wide association study of African Americans." *Hum Mol Genet* **21**(13): 3063-3072.
- 96** . Downs, J. R., M. Clearfield, S. Weis, E. Whitney, D. R. Shapiro, P. A. Beere, . . . A. M. Gotto, Jr. (1998). "Primary prevention of acute coronary events with lovastatin in men and women with average cholesterol levels: results of AFCAPS/TexCAPS. Air Force/Texas Coronary Atherosclerosis Prevention Study." *JAMA* **279**(20): 1615-1622.
- 97** . Duewell, P., H. Kono, K. J. Rayner, C. M. Sirois, G. Vladimer, F. G. Bauernfeind, . . . E. Latz (2010). "NLRP3 inflammasomes are required for atherogenesis and activated by cholesterol crystals." *Nature* **464**(7293): 1357-1361.
- 98** . Dzau, V. and E. Braunwald (1991). "Resolved and unresolved issues in the prevention and treatment of coronary artery disease: a workshop consensus statement." *Am Heart J* **121**(4 Pt 1): 1244-1263.
- 99** . Dzau, V. J., E. M. Antman, H. R. Black, D. L. Hayes, J. E. Manson, J. Plutzky, . . . W. Stevenson (2006). "The cardiovascular disease continuum validated: clinical evidence of improved patient outcomes: part II: Clinical trial evidence (acute coronary syndromes through renal disease) and future directions." *Circulation* **114**(25): 2871-2891.
- 100** . Ehret, G. B., P. B. Munroe, K. M. Rice, M. Bochud, A. D. Johnson, D. I. Chasman, . . . L. Lightstone (2011). "Genetic variants in novel pathways influence blood pressure and cardiovascular disease risk." *Nature* **478**(7367): 103-109.
- 101** . Eichler, E. E., J. Flint, G. Gibson, A. Kong, S. M. Leal, J. H. Moore and J. H. Nadeau (2010). "Missing heritability and strategies for finding the underlying causes of complex disease." *Nat Rev Genet* **11**(6): 446-450.
- 102** . Elliott, P., J. C. Chambers, W. Zhang, R. Clarke, J. C. Hopewell, J. F. Peden, . . . J. S. Kooner (2009). "Genetic Loci associated with C-reactive protein levels and risk of coronary heart disease." *JAMA* **302**(1): 37-48.
- 103** . Ellis, J., E. M. Lange, J. Li, J. Dupuis, J. Baumert, J. D. Walston, . . . L. A. Lange (2014). "Large multiethnic Candidate Gene Study for C-reactive protein levels: identification of a novel association at CD36 in African Americans." *Hum Genet* **133**(8): 985-995.
- 104** . Elston, R. C. and J. Stewart (1971). "A general model for the genetic analysis of pedigree data." *Hum Hered* **21**(6): 523-542.
- 105** . Emerging Risk Factors, C., E. Di Angelantonio, N. Sarwar, P. Perry, S. Kaptoge, K. K. Ray, . . . J. Danesh (2009). "Major lipids, apolipoproteins, and risk of vascular disease." *JAMA* **302**(18): 1993-2000.
- 106** . Emerging Risk Factors, C., S. Kaptoge, E. Di Angelantonio, G. Lowe, M. B. Pepys, S. G. Thompson, . . . J. Danesh (2010). "C-reactive protein concentration and risk of coronary heart disease, stroke, and mortality: an individual participant meta-analysis." *Lancet* **375**(9709): 132-140.
- 107** . Emi, M., D. E. Wilson, P. H. Iverius, L. Wu, A. Hata, R. Hegele, . . . J. M. Lalouel (1990). "Missense mutation (Gly----Glu188) of human lipoprotein lipase imparting functional deficiency." *J Biol Chem* **265**(10): 5910-5916.

- 108** . Endo, A., M. Kuroda and K. Tanzawa (1976). "Competitive inhibition of 3-hydroxy-3-methylglutaryl coenzyme A reductase by ML-236A and ML-236B fungal metabolites, having hypocholesterolemic activity." *FEBS Lett* **72**(2): 323-326.
- 109** . Esko, T., M. Mezzavilla, M. Nelis, C. Borel, T. Debniak, E. Jakkula, . . . P. D'Adamo (2013). "Genetic characterization of northeastern Italian population isolates in the context of broader European genetic diversity." *Eur J Hum Genet* **21**(6): 659-665.
- 110** . Expert Panel on Detection, E. and A. Treatment of High Blood Cholesterol in (2001). "Executive Summary of The Third Report of The National Cholesterol Education Program (NCEP) Expert Panel on Detection, Evaluation, And Treatment of High Blood Cholesterol In Adults (Adult Treatment Panel III)." *JAMA* **285**(19): 2486-2497.
- 111** . Feinleib, M., W. B. Kannel, R. J. Garrison, P. M. McNamara and W. P. Castelli (1975). "The Framingham Offspring Study. Design and preliminary data." *Prev Med* **4**(4): 518-525.
- 112** . Feng, S., D. Liu, X. Zhan, M. K. Wing and G. R. Abecasis (2014). "RAREMETAL: fast and powerful meta-analysis for rare variants." *Bioinformatics*.
- 113** . Ferreira, M. A., J. J. Hottenga, N. M. Warrington, S. E. Medland, G. Willemsen, R. W. Lawrence, . . . D. I. Boomsma (2009). "Sequence variants in three loci influence monocyte counts and erythrocyte volume." *Am J Hum Genet* **85**(5): 745-749.
- 114** . Fischer, M., U. Broeckel, S. Holmer, A. Baessler, C. Hengstenberg, B. Mayer, . . . H. Schunkert (2005). "Distinct heritable patterns of angiographic coronary artery disease in families with myocardial infarction." *Circulation* **111**(7): 855-862.
- 115** . Flannick, J., J. M. Korn, P. Fontanillas, G. B. Grant, E. Banks, M. A. Depristo and D. Altshuler (2012). "Efficiency and power as a function of sequence coverage, SNP array density, and imputation." *PLoS Comput Biol* **8**(7): e1002604.
- 116** . Folsom, A. R., L. E. Chambless, C. M. Ballantyne, J. Coresh, G. Heiss, K. K. Wu, . . . A. R. Sharrett (2006). "An assessment of incremental coronary risk prediction using C-reactive protein and other novel risk markers: the atherosclerosis risk in communities study." *Arch Intern Med* **166**(13): 1368-1373.
- 117** . Fredrickson, D. S. and R. S. Lees (1965). "A System for Phenotyping Hyperlipoproteinemia." *Circulation* **31**: 321-327.
- 118** . Friedewald, W. T., R. I. Levy and D. S. Fredrickson (1972). "Estimation of the concentration of low-density lipoprotein cholesterol in plasma, without use of the preparative ultracentrifuge." *Clin Chem* **18**(6): 499-502.
- 119** . Frikke-Schmidt, R., B. G. Nordestgaard, M. C. Stene, A. A. Sethi, A. T. Remaley, P. Schnohr, . . . A. Tybjaerg-Hansen (2008). "Association of loss-of-function mutations in the ABCA1 gene with high-density lipoprotein cholesterol levels and risk of ischemic heart disease." *JAMA* **299**(21): 2524-2532.
- 120** . Funke, H., A. von Eckardstein, P. H. Pritchard, J. J. Albers, J. J. Kastelein, C. Droste and G. Assmann (1991). "A molecular defect causing fish eye disease: an amino acid exchange in lecithin-cholesterol acyltransferase (LCAT) leads to the selective loss of alpha-LCAT activity." *Proc Natl Acad Sci U S A* **88**(11): 4855-4859.
- 121** . Gambaro, G., T. Yabarek, M. S. Graziani, A. Gemelli, C. Abaterusso, A. C. Frigo, . . . I. S. Group (2010). "Prevalence of CKD in northeastern Italy: results of the INCIPE study and comparison with NHANES." *Clin J Am Soc Nephrol* **5**(11): 1946-1953.
- 122** . Ganesh, S. K., N. A. Zakai, F. J. van Rooij, N. Soranzo, A. V. Smith, M. A. Nalls, . . . J. P. Lin (2009). "Multiple loci influence erythrocyte phenotypes in the CHARGE Consortium." *Nat Genet* **41**(11): 1191-1198.
- 123** . Garcia, C. K., K. Wilund, M. Arca, G. Zuliani, R. Fellin, M. Maioli, . . . H. H. Hobbs (2001). "Autosomal recessive hypercholesterolemia caused by mutations in a putative LDL receptor adaptor protein." *Science* **292**(5520): 1394-1398.
- 124** . Garner, C., T. Tatu, J. E. Reittie, T. Littlewood, J. Darley, S. Cervino, . . . S. L. Thein (2000). "Genetic influences on F cells and other hematologic variables: a twin heritability study." *Blood* **95**(1): 342-346.

- 125** . Gieger, C., A. Radhakrishnan, A. Cvejic, W. Tang, E. Porcu, G. Pistis, . . . N. Soranzo (2011). "New gene functions in megakaryopoiesis and platelet formation." Nature **480**(7376): 201-208.
- 126** . Glessner, J. T., K. Wang, G. Cai, O. Korvatska, C. E. Kim, S. Wood, . . . H. Hakonarson (2009). "Autism genome-wide copy number variation reveals ubiquitin and neuronal genes." Nature **459**(7246): 569-573.
- 127** . Global Lipids Genetics, C., C. J. Willer, E. M. Schmidt, S. Sengupta, G. M. Peloso, S. Gustafsson, . . . G. R. Abecasis (2013). "Discovery and refinement of loci associated with lipid levels." Nat Genet **45**(11): 1274-1283.
- 128** . Glud, T., E. B. Schmidt, S. D. Kristensen and T. Arnfred (1986). "Platelet number and volume during myocardial infarction in relation to infarct size." Acta Med Scand **220**(5): 401-405.
- 129** . Goate, A., M. C. Chartier-Harlin, M. Mullan, J. Brown, F. Crawford, L. Fidani, . . . et al. (1991). "Segregation of a missense mutation in the amyloid precursor protein gene with familial Alzheimer's disease." Nature **349**(6311): 704-706.
- 130** . Goldbourt, U., S. Yaari and J. H. Medalie (1997). "Isolated low HDL cholesterol as a risk factor for coronary heart disease mortality. A 21-year follow-up of 8000 men." Arterioscler Thromb Vasc Biol **17**(1): 107-113.
- 131** . Golding, J., M. Pembrey and R. Jones (2001). "ALSPAC--the Avon Longitudinal Study of Parents and Children. I. Study methodology." Paediatr Perinat Epidemiol **15**(1): 74-87.
- 132** . Golding, J., M. Pembrey, R. Jones and A. S. Team (2001). "ALSPAC--the Avon Longitudinal Study of Parents and Children. I. Study methodology." Paediatr Perinat Epidemiol **15**(1): 74-87.
- 133** . Goldstein, D. B. (2009). "Common genetic variation and human traits." N Engl J Med **360**(17): 1696-1698.
- 134** . Goldstein, D. B., A. Allen, J. Keebler, E. H. Margulies, S. Petrou, S. Petrovski and S. Sunyaev (2013). "Sequencing studies in human genetics: design and interpretation." Nat Rev Genet **14**(7): 460-470.
- 135** . Goode, E. L., S. S. Cherny, J. C. Christian, G. P. Jarvik and M. de Andrade (2007). "Heritability of longitudinal measures of body mass index and lipid and lipoprotein levels in aging twins." Twin Res Hum Genet **10**(5): 703-711.
- 136** . Gordon, D. J., J. L. Probstfield, R. J. Garrison, J. D. Neaton, W. P. Castelli, J. D. Knoke, . . . H. A. Tyroler (1989). "High-density lipoprotein cholesterol and cardiovascular disease. Four prospective American studies." Circulation **79**(1): 8-15.
- 137** . Gordon, T., W. P. Castelli, M. C. Hjortland, W. B. Kannel and T. R. Dawber (1977). "High density lipoprotein as a protective factor against coronary heart disease. The Framingham Study." Am J Med **62**(5): 707-714.
- 138** . Graham, I., D. Atar, K. Borch-Johnsen, G. Boysen, G. Burell, R. Cifkova, . . . G. European Society of Cardiology Committee for Practice (2007). "European guidelines on cardiovascular disease prevention in clinical practice: executive summary: Fourth Joint Task Force of the European Society of Cardiology and Other Societies on Cardiovascular Disease Prevention in Clinical Practice (Constituted by representatives of nine societies and by invited experts)." Eur Heart J **28**(19): 2375-2414.
- 139** . Greenburg, A. G. (1996). "Pathophysiology of anemia." Am J Med **101**(2A): 7S-11S.
- 140** . Haase, C. L., A. Tybjaerg-Hansen, A. A. Qayyum, J. Schou, B. G. Nordestgaard and R. Frikke-Schmidt (2012). "LCAT, HDL cholesterol and ischemic cardiovascular disease: a Mendelian randomization study of HDL cholesterol in 54,500 individuals." J Clin Endocrinol Metab **97**(2): E248-256.
- 141** . Haines, J. L., M. A. Hauser, S. Schmidt, W. K. Scott, L. M. Olson, P. Gallins, . . . M. A. Pericak-Vance (2005). "Complement factor H variant increases the risk of age-related macular degeneration." Science **308**(5720): 419-421.
- 142** . Hardison, R. C. and G. A. Blobel (2013). "Genetics. GWAS to therapy by genome edits?" Science **342**(6155): 206-207.

- 143** . Harrow, J., A. Frankish, J. M. Gonzalez, E. Tapanari, M. Diekhans, F. Kokocinski, . . . T. J. Hubbard (2012). "GENCODE: the reference human genome annotation for The ENCODE Project." Genome Res **22**(9): 1760-1774.
- 144** . Hays, J., J. R. Hunt, F. A. Hubbell, G. L. Anderson, M. Limacher, C. Allen and J. E. Rossouw (2003). "The Women's Health Initiative recruitment methods and results." Ann Epidemiol **13**(9 Suppl): S18-77.
- 145** . He, Z., B. J. O'Roak, J. D. Smith, G. Wang, S. Hooker, R. L. Santos-Cortez, . . . S. M. Leal (2014). "Rare-variant extensions of the transmission disequilibrium test: application to autism exome sequence data." Am J Hum Genet **94**(1): 33-46.
- 146** . Heart Protection Study Collaborative, G. (2002). "MRC/BHF Heart Protection Study of cholesterol lowering with simvastatin in 20,536 high-risk individuals: a randomised placebo-controlled trial." Lancet **360**(9326): 7-22.
- 147** . Hegele, R. A., J. A. Little and P. W. Connelly (1991). "Compound heterozygosity for mutant hepatic lipase in familial hepatic lipase deficiency." Biochem Biophys Res Commun **179**(1): 78-84.
- 148** . Heid, I. M., E. Boes, M. Muller, B. Kollerits, C. Lamina, S. Coassin, . . . F. Kronenberg (2008). "Genome-wide association analysis of high-density lipoprotein cholesterol in the population-based KORA study sheds new light on intergenic regions." Circ Cardiovasc Genet **1**(1): 10-20.
- 149** . Helfand, M., D. I. Buckley, M. Freeman, R. Fu, K. Rogers, C. Fleming and L. L. Humphrey (2009). "Emerging risk factors for coronary heart disease: a summary of systematic reviews conducted for the U.S. Preventive Services Task Force." Ann Intern Med **151**(7): 496-507.
- 150** . Hemani, G., J. Yang, A. Vinkhuyzen, J. E. Powell, G. Willemsen, J. J. Hottenga, . . . P. M. Visscher (2013). "Inference of the genetic architecture underlying BMI and height with the use of 20,240 sibling pairs." Am J Hum Genet **93**(5): 865-875.
- 151** . Hendra, T. J., G. A. Oswald and J. S. Yudkin (1988). "Increased mean platelet volume after acute myocardial infarction relates to diabetes and to cardiac failure." Diabetes Res Clin Pract **5**(1): 63-69.
- 152** . Hindorff, L. A., P. Sethupathy, H. A. Junkins, E. M. Ramos, J. P. Mehta, F. S. Collins and T. A. Manolio (2009). "Potential etiologic and functional implications of genome-wide association loci for human diseases and traits." Proc Natl Acad Sci U S A **106**(23): 9362-9367.
- 153** . Hirschhorn, J. N. and M. J. Daly (2005). "Genome-wide association studies for common diseases and complex traits." Nat Rev Genet **6**(2): 95-108.
- 154** . Hiura, Y., C. S. Shen, Y. Kokubo, T. Okamura, T. Morisaki, H. Tomoike, . . . N. Iwai (2009). "Identification of genetic markers associated with high-density lipoprotein-cholesterol by genome-wide screening in a Japanese population: the Suita study." Circ J **73**(6): 1119-1126.
- 155** . Hoffman, M., A. Blum, R. Baruch, E. Kaplan and M. Benjamin (2004). "Leukocytes and coronary heart disease." Atherosclerosis **172**(1): 1-6.
- 156** . Holm, H., D. F. Gudbjartsson, P. Sulem, G. Masson, H. T. Helgadóttir, C. Zanon, . . . K. Stefansson (2011). "A rare variant in MYH6 is associated with high risk of sick sinus syndrome." Nat Genet **43**(4): 316-320.
- 157** . Holmen, O. L., H. Zhang, Y. Fan, D. H. Hovelson, E. M. Schmidt, W. Zhou, . . . C. J. Willer (2014). "Systematic evaluation of coding variation identifies a candidate causal variant in TM6SF2 influencing total cholesterol and myocardial infarction risk." Nat Genet **46**(4): 345-351.
- 158** . Holmen, O. L., H. Zhang, W. Zhou, E. Schmidt, D. H. Hovelson, A. Langhammer, . . . C. J. Willer (2014). "No large-effect low-frequency coding variation found for myocardial infarction." Hum Mol Genet **23**(17): 4721-4728.
- 159** . Howie, B., C. Fuchsberger, M. Stephens, J. Marchini and G. R. Abecasis (2012). "Fast and accurate genotype imputation in genome-wide association studies through pre-phasing." Nat Genet **44**(8): 955-959.
- 160** . Howie, B., J. Marchini and M. Stephens (2011). "Genotype imputation with thousands of genomes." G3 (Bethesda) **1**(6): 457-470.
- 161** . Howie, B. N., P. Donnelly and J. Marchini (2009). "A flexible and accurate genotype imputation method for the next generation of genome-wide association studies." PLoS Genet **5**(6): e1000529.

- 162** . Huang, J., D. Ellinghaus, A. Franke, B. Howie and Y. Li (2012). "1000 Genomes-based imputation identifies novel and refined associations for the Wellcome Trust Case Control Consortium phase 1 Data." Eur J Hum Genet.
- 163** . Huang, J., B. Howie, S. McCarthy, Y. Memari, K. Walter, J. Min, . . . N. Soranzo (2015). "A reference panel of 3,781 genomes from the UK10K Project increases imputation performance of low frequency variants." Nature Communications (Under peer review).
- 164** . Huang, J., A. D. Johnson and C. J. O'Donnell (2011). "PRIME: a method for characterization and evaluation of pleiotropic regions from multiple genome-wide association studies." Bioinformatics **27**(9): 1201-1206.
- 165** . Huang, J., R. H. Perlis, P. H. Lee, A. J. Rush, M. Fava, G. S. Sachs, . . . J. W. Smoller (2010). "Cross-disorder genomewide analysis of schizophrenia, bipolar disorder, and depression." Am J Psychiatry **167**(10): 1254-1263.
- 166** . Hunink, M. G., L. Goldman, A. N. Tosteson, M. A. Mittleman, P. A. Goldman, L. W. Williams, . . . M. C. Weinstein (1997). "The recent decline in mortality from coronary heart disease, 1980-1990. The effect of secular trends in risk factors and treatment." JAMA **277**(7): 535-542.
- 167** . Hunt, K. A., V. Mistry, N. A. Bockett, T. Ahmad, M. Ban, J. N. Barker, . . . D. A. van Heel (2013). "Negligible impact of rare autoimmune-locus coding-region variants on missing heritability." Nature **498**(7453): 232-235.
- 168** . Ibanez, B., G. Vilahur and J. J. Badimon (2007). "Plaque progression and regression in atherothrombosis." J Thromb Haemost **5 Suppl 1**: 292-299.
- 169** . Idaghdour, Y., J. Quinlan, J. P. Goulet, J. Berghout, E. Gbeha, V. Bruat, . . . P. Awadalla (2012). "Evidence for additive and interaction effects of host genotype and infection in malaria." Proc Natl Acad Sci U S A **109**(42): 16786-16793.
- 170** . Igl, W., A. Johansson, J. F. Wilson, S. H. Wild, O. Polasek, C. Hayward, . . . E. Consortium (2010). "Modeling of environmental effects in genome-wide association studies identifies SLC2A2 and HP as novel loci influencing serum cholesterol levels." PLoS Genet **6**(1): e1000798.
- 171** . Ingelsson, E., E. J. Schaefer, J. H. Contois, J. R. McNamara, L. Sullivan, M. J. Keyes, . . . R. S. Vasan (2007). "Clinical utility of different lipid measures for prediction of coronary heart disease in men and women." JAMA **298**(7): 776-785.
- 172** . Interleukin-6 Receptor Mendelian Randomisation Analysis, C., A. D. Hingorani and J. P. Casas (2012). "The interleukin-6 receptor as a target for prevention of coronary heart disease: a mendelian randomisation analysis." Lancet **379**(9822): 1214-1224.
- 173** . International HapMap, C., D. M. Altshuler, R. A. Gibbs, L. Peltonen, D. M. Altshuler, R. A. Gibbs, . . . J. E. McEwen (2010). "Integrating common and rare genetic variation in diverse human populations." Nature **467**(7311): 52-58.
- 174** . International HapMap, C., K. A. Frazer, D. G. Ballinger, D. R. Cox, D. A. Hinds, L. L. Stuve, . . . J. Stewart (2007). "A second generation human haplotype map of over 3.1 million SNPs." Nature **449**(7164): 851-861.
- 175** . International Schizophrenia, C. (2008). "Rare chromosomal deletions and duplications increase risk of schizophrenia." Nature **455**(7210): 237-241.
- 176** . International Schizophrenia, C., S. M. Purcell, N. R. Wray, J. L. Stone, P. M. Visscher, M. C. O'Donovan, . . . P. Sklar (2009). "Common polygenic variation contributes to risk of schizophrenia and bipolar disorder." Nature **460**(7256): 748-752.
- 177** . Jallow, M., Y. Y. Teo, K. S. Small, K. A. Rockett, P. Deloukas, T. G. Clark, . . . N. Malaria Genomic Epidemiology (2009). "Genome-wide and fine-resolution association analysis of malaria in West Africa." Nat Genet **41**(6): 657-665.
- 178** . Jewett, E. M., M. Zawistowski, N. A. Rosenberg and S. Zollner (2012). "A coalescent model for genotype imputation." Genetics **191**(4): 1239-1255.
- 179** . Johannsen, T. H., P. R. Kamstrup, R. V. Andersen, G. B. Jensen, H. Sillesen, A. Tybjaerg-Hansen and B. G. Nordestgaard (2009). "Hepatic lipase, genetically elevated high-density lipoprotein, and risk of ischemic cardiovascular disease." J Clin Endocrinol Metab **94**(4): 1264-1273.

- 180** . Johansen, C. T., J. Wang, M. B. Lanktree, H. Cao, A. D. McIntyre, M. R. Ban, . . . R. A. Hegele (2010). "Excess of rare variants in genes identified by genome-wide association study of hypertriglyceridemia." Nat Genet **42**(8): 684-687.
- 181** . Jones, B., E. L. Jones, S. A. Bonney, H. N. Patel, A. R. Mensenkamp, S. Eichenbaum-Voline, . . . C. C. Shoulders (2003). "Mutations in a Sar1 GTPase of COPII vesicles are associated with lipid absorption disorders." Nat Genet **34**(1): 29-31.
- 182** . Jorgensen, A. B., R. Frikke-Schmidt, B. G. Nordestgaard and A. Tybjaerg-Hansen (2014). "Loss-of-Function Mutations in APOC3 and Risk of Ischemic Vascular Disease." N Engl J Med **371**(1): 32-41.
- 183** . Kamatani, Y., K. Matsuda, Y. Okada, M. Kubo, N. Hosono, Y. Daigo, . . . N. Kamatani (2010). "Genome-wide association study of hematological and biochemical traits in a Japanese population." Nat Genet **42**(3): 210-215.
- 184** . Kannel, W. B., K. Anderson and P. W. Wilson (1992). "White blood cell count and cardiovascular disease. Insights from the Framingham Study." JAMA **267**(9): 1253-1256.
- 185** . Kannel, W. B., T. R. Dawber, G. D. Friedman, W. E. Glennon and P. M. McNamara (1964). "Risk Factors in Coronary Heart Disease. An Evaluation of Several Serum Lipids as Predictors of Coronary Heart Disease; the Framingham Study." Ann Intern Med **61**: 888-899.
- 186** . Kannel, W. B., T. R. Dawber, A. Kagan, N. Revotskie and J. Stokes, 3rd (1961). "Factors of risk in the development of coronary heart disease--six year follow-up experience. The Framingham Study." Ann Intern Med **55**: 33-50.
- 187** . Kannel, W. B., T. R. Dawber and D. L. McGee (1980). "Perspectives on systolic hypertension. The Framingham study." Circulation **61**(6): 1179-1182.
- 188** . Kannel, W. B., R. S. Vasan, M. J. Keyes, L. M. Sullivan and S. J. Robins (2008). "Usefulness of the triglyceride-high-density lipoprotein versus the cholesterol-high-density lipoprotein ratio for predicting insulin resistance and cardiometabolic risk (from the Framingham Offspring Cohort)." Am J Cardiol **101**(4): 497-501.
- 189** . Kannel, W. B., P. A. Wolf, W. P. Castelli and R. B. D'Agostino (1987). "Fibrinogen and risk of cardiovascular disease. The Framingham Study." JAMA **258**(9): 1183-1186.
- 190** . Kathiresan, S., A. K. Manning, S. Demissie, R. B. D'Agostino, A. Surti, C. Guiducci, . . . L. A. Cupples (2007). "A genome-wide association study for blood lipid phenotypes in the Framingham Heart Study." BMC Med Genet **8 Suppl 1**: S17.
- 191** . Kathiresan, S., O. Melander, C. Guiducci, A. Surti, N. P. Burt, M. J. Rieder, . . . M. Orho-Melander (2008). "Six new loci associated with blood low-density lipoprotein cholesterol, high-density lipoprotein cholesterol or triglycerides in humans." Nat Genet **40**(2): 189-197.
- 192** . Kathiresan, S. and D. Srivastava (2012). "Genetics of human cardiovascular disease." Cell **148**(6): 1242-1257.
- 193** . Kathiresan, S., C. J. Willer, G. M. Peloso, S. Demissie, K. Musunuru, E. E. Schadt, . . . L. A. Cupples (2009). "Common variants at 30 loci contribute to polygenic dyslipidemia." Nat Genet **41**(1): 56-65.
- 194** . Keller, M., D. Schleinitz, J. Forster, A. Tonjes, Y. Bottcher, A. Fischer-Rosinsky, . . . P. Kovacs (2013). "THOC5: a novel gene involved in HDL-cholesterol metabolism." J Lipid Res **54**(11): 3170-3176.
- 195** . Keller, M. F., A. P. Reiner, Y. Okada, F. J. van Rooij, A. D. Johnson, M. H. Chen, . . . G. BioBank Japan Project Working (2014). "Trans-ethnic meta-analysis of white blood cell phenotypes." Hum Mol Genet **23**(25): 6944-6960.
- 196** . Kerem, B., J. M. Rommens, J. A. Buchanan, D. Markiewicz, T. K. Cox, A. Chakravarti, . . . L. C. Tsui (1989). "Identification of the cystic fibrosis gene: genetic analysis." Science **245**(4922): 1073-1080.
- 197** . Keskin, O., R. E. Ulusoy, M. Kalemoglu, M. H. Us, I. Yildirim, O. Tarcin, . . . N. Ardic (2004). "White blood cell count and C-reactive protein predict short-term prognosis in acute myocardial infarction." J Int Med Res **32**(6): 646-654.

- 198** . Kettunen, J., T. Tukiainen, A. P. Sarin, A. Ortega-Alonso, E. Tikkanen, L. P. Lyytikäinen, . . . S. Ripatti (2012). "Genome-wide association study identifies multiple loci influencing human serum metabolite levels." Nat Genet **44**(3): 269-276.
- 199** . Kim, S., S. Swaminathan, L. Shen, S. L. Risacher, K. Nho, T. Foroud, . . . I. Alzheimer's Disease Neuroimaging (2011). "Genome-wide association study of CSF biomarkers Abeta1-42, t-tau, and p-tau181p in the ADNI cohort." Neurology **76**(1): 69-79.
- 200** . Kim, S. Y., J. P. Guevara, K. M. Kim, H. K. Choi, D. F. Heitjan and D. A. Albert (2010). "Hyperuricemia and coronary heart disease: a systematic review and meta-analysis." Arthritis Care Res (Hoboken) **62**(2): 170-180.
- 201** . Kim, Y. J., M. J. Go, C. Hu, C. B. Hong, Y. K. Kim, J. Y. Lee, . . . Y. S. Cho (2011). "Large-scale genome-wide association studies in East Asians identify new genetic loci influencing metabolic traits." Nat Genet **43**(10): 990-995.
- 202** . Klein, R. J., C. Zeiss, E. Y. Chew, J. Y. Tsai, R. S. Sackler, C. Haynes, . . . J. Hoh (2005). "Complement factor H polymorphism in age-related macular degeneration." Science **308**(5720): 385-389.
- 203** . Koenig, W., H. Lowel, J. Baumert and C. Meisinger (2004). "C-reactive protein modulates risk prediction based on the Framingham Score: implications for future risk assessment: results from a large cohort study in southern Germany." Circulation **109**(11): 1349-1353.
- 204** . Kong, M. and C. Lee (2013). "Genetic associations with C-reactive protein level and white blood cell count in the KARE study." Int J Immunogenet **40**(2): 120-125.
- 205** . Kooner, J. S., J. C. Chambers, C. A. Aguilar-Salinas, D. A. Hinds, C. L. Hyde, G. R. Warnes, . . . J. F. Thompson (2008). "Genome-wide scan identifies variation in MLXIPL associated with plasma triglycerides." Nat Genet **40**(2): 149-151.
- 206** . Kuivenhoven, J. A. and R. A. Hegele (2014). "Mining the genome for lipid genes." Biochim Biophys Acta **1842**(10): 1993-2009.
- 207** . Kuller, L. H. (1976). "Epidemiology of cardiovascular diseases: current perspectives." Am J Epidemiol **104**(4): 425-496.
- 208** . Kullo, I. J., K. Ding, H. Jouni, C. Y. Smith and C. G. Chute (2010). "A genome-wide association study of red blood cell traits using the electronic medical record." PLoS One **5**(9).
- 209** . Kuroda, M., Y. Tsujita, K. Tanzawa and A. Endo (1979). "Hypolipidemic effects in monkeys of ML-236B, a competitive inhibitor of 3-hydroxy-3-methylglutaryl coenzyme A reductase." Lipids **14**(6): 585-589.
- 210** . Kwiatkowski, D. P. (2005). "How malaria has affected the human genome and what human genetics can teach us about malaria." Am J Hum Genet **77**(2): 171-192.
- 211** . Labreuche, J., P. J. Touboul and P. Amarenco (2009). "Plasma triglyceride levels and risk of stroke and carotid atherosclerosis: a systematic review of the epidemiological studies." Atherosclerosis **203**(2): 331-345.
- 212** . Ladouceur, M., Z. Dastani, Y. S. Aulchenko, C. M. Greenwood and J. B. Richards (2012). "The empirical power of rare variant association methods: results from sanger sequencing in 1,998 individuals." PLoS Genet **8**(2): e1002496.
- 213** . Lage, K., E. O. Karlberg, Z. M. Storling, P. I. Olason, A. G. Pedersen, O. Rigina, . . . S. Brunak (2007). "A human phenome-interactome network of protein complexes implicated in genetic disorders." Nat Biotechnol **25**(3): 309-316.
- 214** . LaMonte, G., N. Philip, J. Reardon, J. R. Lacsina, W. Majoros, L. Chapman, . . . J. T. Chi (2012). "Translocation of sickle cell erythrocyte microRNAs into Plasmodium falciparum inhibits parasite translation and contributes to malaria resistance." Cell Host Microbe **12**(2): 187-199.
- 215** . Lander, E. S. (1996). "The new genomics: global views of biology." Science **274**(5287): 536-539.
- 216** . Lander, E. S. and P. Green (1987). "Construction of multilocus genetic linkage maps in humans." Proc Natl Acad Sci U S A **84**(8): 2363-2367.

- 217** . Lander, E. S., L. M. Linton, B. Birren, C. Nusbaum, M. C. Zody, J. Baldwin, . . . C. International Human Genome Sequencing (2001). "Initial sequencing and analysis of the human genome." Nature **409**(6822): 860-921.
- 218** . Langaee, T. and M. Ronaghi (2005). "Genetic variation analyses by Pyrosequencing." Mutat Res **573**(1-2): 96-102.
- 219** . Lange, L. A., Y. Hu, H. Zhang, C. Xue, E. M. Schmidt, Z. Z. Tang, . . . N. G. O. E. S. Project (2014). "Whole-exome sequencing identifies rare and low-frequency coding variants associated with LDL cholesterol." Am J Hum Genet **94**(2): 233-245.
- 220** . Lango Allen, H., K. Estrada, G. Lettre, S. I. Berndt, M. N. Weedon, F. Rivadeneira, . . . J. N. Hirschhorn (2010). "Hundreds of variants clustered in genomic loci and biological pathways affect human height." Nature **467**(7317): 832-838.
- 221** . Lanzara, C., A. d'Adamo and M. Montico (2015). "Use of an Italian isolated population for studying complex diseases. The Carlantino project: study design and preliminary results. ." Slovenian J Pub Health(in press).
- 222** . Lavie, C. J. and R. V. Milani (2003). "Obesity and cardiovascular disease: the hippocrates paradox?" J Am Coll Cardiol **42**(4): 677-679.
- 223** . Lawlor, D. A., R. M. Harbord, N. J. Timpson, G. D. Lowe, A. Rumley, T. R. Gaunt, . . . G. D. Smith (2008). "The association of C-reactive protein and CRP genotype with coronary heart disease: findings from five studies with 4,610 cases amongst 18,637 participants." PLoS One **3**(8): e3011.
- 224** . Le, S. Q. and R. Durbin (2011). "SNP detection and genotyping from low-coverage sequencing data on multiple diploid samples." Genome Res **21**(6): 952-960.
- 225** . Lee, S., M. J. Emond, M. J. Bamshad, K. C. Barnes, M. J. Rieder, D. A. Nickerson, . . . X. Lin (2012). "Optimal unified approach for rare-variant association testing with application to small-sample case-control whole-exome sequencing studies." Am J Hum Genet **91**(2): 224-237.
- 226** . Lee, S., M. C. Wu and X. Lin (2012). "Optimal tests for rare variant effects in sequencing association studies." Biostatistics **13**(4): 762-775.
- 227** . Lehrman, M. A., J. L. Goldstein, M. S. Brown, D. W. Russell and W. J. Schneider (1985). "Internalization-defective LDL receptors produced by genes with nonsense and frameshift mutations that truncate the cytoplasmic domain." Cell **41**(3): 735-743.
- 228** . Lemieux, I., B. Lamarche, C. Couillard, A. Pascot, B. Cantin, J. Bergeron, . . . J. P. Despres (2001). "Total cholesterol/HDL cholesterol ratio vs LDL cholesterol/HDL cholesterol ratio as indices of ischemic heart disease risk in men: the Quebec Cardiovascular Study." Arch Intern Med **161**(22): 2685-2692.
- 229** . Lewis, G. F. and D. J. Rader (2005). "New insights into the regulation of HDL metabolism and reverse cholesterol transport." Circ Res **96**(12): 1221-1232.
- 230** . Li, B. 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**(3): 311-321.
- 231** . Li, H., B. Handsaker, A. Wysoker, T. Fennell, J. Ruan, N. Homer, . . . S. Genome Project Data Processing (2009). "The Sequence Alignment/Map format and SAMtools." Bioinformatics **25**(16): 2078-2079.
- 232** . Li, J., J. T. Glessner, H. Zhang, C. Hou, Z. Wei, J. P. Bradfield, . . . P. M. Sleiman (2013). "GWAS of blood cell traits identifies novel associated loci and epistatic interactions in Caucasian and African-American children." Hum Mol Genet **22**(7): 1457-1464.
- 233** . Li, N. and M. Stephens (2003). "Modeling linkage disequilibrium and identifying recombination hotspots using single-nucleotide polymorphism data." Genetics **165**(4): 2213-2233.
- 234** . Li, Y., C. Sidore, H. M. Kang, M. Boehnke and G. R. Abecasis (2011). "Low-coverage sequencing: implications for design of complex trait association studies." Genome Res **21**(6): 940-951.
- 235** . Li, Y., C. Willer, S. Sanna and G. Abecasis (2009). "Genotype imputation." Annu Rev Genomics Hum Genet **10**: 387-406.
- 236** . Libby, P. (2002). "Inflammation in atherosclerosis." Nature **420**(6917): 868-874.

- 237** . Lim, E. T., P. Wurtz, A. S. Havulinna, P. Palta, T. Tukiainen, K. Rehnstrom, . . . P. Sequencing Initiative Suomi (2014). "Distribution and medical impact of loss-of-function variants in the Finnish founder population." *PLoS Genet* **10**(7): e1004494.
- 238** . Lin, D. Y. and Z. Z. Tang (2011). "A general framework for detecting disease associations with rare variants in sequencing studies." *Am J Hum Genet* **89**(3): 354-367.
- 239** . Lin, J. P., C. J. O'Donnell, L. Jin, C. Fox, Q. Yang and L. A. Cupples (2007). "Evidence for linkage of red blood cell size and count: genome-wide scans in the Framingham Heart Study." *Am J Hematol* **82**(7): 605-610.
- 240** . Linsel-Nitschke, P., A. Gotz, J. Erdmann, I. Braenne, P. Braund, C. Hengstenberg, . . . C. Cardiogenics (2008). "Lifelong reduction of LDL-cholesterol related to a common variant in the LDL-receptor gene decreases the risk of coronary artery disease--a Mendelian Randomisation study." *PLoS One* **3**(8): e2986.
- 241** . Liu, D. J. and S. M. Leal (2012). "Estimating genetic effects and quantifying missing heritability explained by identified rare-variant associations." *Am J Hum Genet* **91**(4): 585-596.
- 242** . Liu, D. J., G. M. Peloso, X. Zhan, O. L. Holmen, M. Zawistowski, S. Feng, . . . G. R. Abecasis (2014). "Meta-analysis of gene-level tests for rare variant association." *Nat Genet* **46**(2): 200-204.
- 243** . Liuzzo, G., L. M. Biasucci, J. R. Gallimore, R. L. Grillo, A. G. Rebuzzi, M. B. Pepys and A. Maseri (1994). "The prognostic value of C-reactive protein and serum amyloid a protein in severe unstable angina." *N Engl J Med* **331**(7): 417-424.
- 244** . Lloyd-Jones, D. M., K. Liu, L. Tian and P. Greenland (2006). "Narrative review: Assessment of C-reactive protein in risk prediction for cardiovascular disease." *Ann Intern Med* **145**(1): 35-42.
- 245** . Lusis, A. J. and P. Pajukanta (2008). "A treasure trove for lipoprotein biology." *Nat Genet* **40**(2): 129-130.
- 246** . Ma, L., J. Yang, H. B. Runesha, T. Tanaka, L. Ferrucci, S. Bandinelli and Y. Da (2010). "Genome-wide association analysis of total cholesterol and high-density lipoprotein cholesterol levels using the Framingham heart study data." *BMC Med Genet* **11**: 55.
- 247** . MacDonald, M. E., A. Novelletto, C. Lin, D. Tagle, G. Barnes, G. Bates, . . . et al. (1992). "The Huntington's disease candidate region exhibits many different haplotypes." *Nat Genet* **1**(2): 99-103.
- 248** . MacGregor, A. J., J. R. Gallimore, T. D. Spector and M. B. Pepys (2004). "Genetic effects on baseline values of C-reactive protein and serum amyloid a protein: a comparison of monozygotic and dizygotic twins." *Clin Chem* **50**(1): 130-134.
- 249** . Magi, R. and A. P. Morris (2010). "GWAMA: software for genome-wide association meta-analysis." *BMC Bioinformatics* **11**: 288.
- 250** . Malik, I., J. Danesh, P. Whincup, V. Bhatia, O. Papacosta, M. Walker, . . . D. Haskard (2001). "Soluble adhesion molecules and prediction of coronary heart disease: a prospective study and meta-analysis." *Lancet* **358**(9286): 971-976.
- 251** . Maller, J. B., G. McVean, J. Byrnes, D. Vukcevic, K. Palin, Z. Su, . . . P. Donnelly (2012). "Bayesian refinement of association signals for 14 loci in 3 common diseases." *Nat Genet* **44**(12): 1294-1301.
- 252** . Manolio, T. A., F. S. Collins, N. J. Cox, D. B. Goldstein, L. A. Hindorff, D. J. Hunter, . . . P. M. Visscher (2009). "Finding the missing heritability of complex diseases." *Nature* **461**(7265): 747-753.
- 253** . Marcais, C., B. Verges, S. Charriere, V. Pruneta, M. Merlin, S. Billon, . . . P. Moulin (2005). "Apoa5 Q139X truncation predisposes to late-onset hyperchylomicronemia due to lipoprotein lipase impairment." *J Clin Invest* **115**(10): 2862-2869.
- 254** . Marchini, J., B. Howie, S. Myers, G. McVean and P. Donnelly (2007). "A new multipoint method for genome-wide association studies by imputation of genotypes." *Nat Genet* **39**(7): 906-913.
- 255** . Mardis, E. R. (2008). "The impact of next-generation sequencing technology on genetics." *Trends Genet* **24**(3): 133-141.
- 256** . Marduel, M., K. Ouguerram, V. Serre, D. Bonnefont-Rousselot, A. Marques-Pinheiro, K. Erik Berge, . . . M. Varret (2013). "Description of a large family with autosomal dominant hypercholesterolemia associated with the APOE p.Leu167del mutation." *Hum Mutat* **34**(1): 83-87.

- 257** . Margolis, K. L., J. E. Manson, P. Greenland, R. J. Rodabough, P. F. Bray, M. Safford, . . . G. Women's Health Initiative Research (2005). "Leukocyte count as a predictor of cardiovascular events and mortality in postmenopausal women: the Women's Health Initiative Observational Study." Arch Intern Med **165**(5): 500-508.
- 258** . Masicampo, E. J. and D. R. Lalande (2012). "A peculiar prevalence of p values just below .05." Q J Exp Psychol (Hove) **65**(11): 2271-2279.
- 259** . Massberg, S., C. Schulz and M. Gawaz (2003). "Role of platelets in the pathophysiology of acute coronary syndrome." Semin Vasc Med **3**(2): 147-162.
- 260** . Maurano, M. T., R. Humbert, E. Rynes, R. E. Thurman, E. Haugen, H. Wang, . . . J. A. Stamatoyannopoulos (2012). "Systematic localization of common disease-associated variation in regulatory DNA." Science **337**(6099): 1190-1195.
- 261** . McCarthy, M. I., G. R. Abecasis, L. R. Cardon, D. B. Goldstein, J. Little, J. P. Ioannidis and J. N. Hirschhorn (2008). "Genome-wide association studies for complex traits: consensus, uncertainty and challenges." Nat Rev Genet **9**(5): 356-369.
- 262** . McCarthy, M. I. and E. Zeggini (2009). "Genome-wide association studies in type 2 diabetes." Curr Diab Rep **9**(2): 164-171.
- 263** . McLaren, C. E., J. C. Barton, V. R. Gordeuk, L. Wu, P. C. Adams, D. M. Reboussin, . . . I. Iron Overload Screening Study Research (2007). "Determinants and characteristics of mean corpuscular volume and hemoglobin concentration in white HFE C282Y homozygotes in the hemochromatosis and iron overload screening study." Am J Hematol **82**(10): 898-905.
- 264** . McLaren, C. E., C. P. Garner, C. C. Constantine, S. McLachlan, C. D. Vulpe, B. M. Snively, . . . G. D. McLaren (2011). "Genome-wide association study identifies genetic loci associated with iron deficiency." PLoS One **6**(3): e17390.
- 265** . McLaren, W., B. Pritchard, D. Rios, Y. Chen, P. Flicek and F. Cunningham (2010). "Deriving the consequences of genomic variants with the Ensembl API and SNP Effect Predictor." Bioinformatics **26**(16): 2069-2070.
- 266** . McMorrnan, B. J., G. Burgio and S. J. Foote (2013). "New insights into the protective power of platelets in malaria infection." Commun Integr Biol **6**(3): e23653.
- 267** . Meisinger, C., H. Prokisch, C. Gieger, N. Soranzo, D. Mehta, D. Rosskopf, . . . A. Doring (2009). "A genome-wide association study identifies three loci associated with mean platelet volume." Am J Hum Genet **84**(1): 66-71.
- 268** . Melander, O., C. Newton-Cheh, P. Almgren, B. Hedblad, G. Berglund, G. Engstrom, . . . T. J. Wang (2009). "Novel and conventional biomarkers for prediction of incident cardiovascular events in the community." JAMA **302**(1): 49-57.
- 269** . Menzel, S., C. Garner, I. Gut, F. Matsuda, M. Yamaguchi, S. Heath, . . . S. L. Thein (2007). "A QTL influencing F cell production maps to a gene encoding a zinc-finger protein on chromosome 2p15." Nat Genet **39**(10): 1197-1199.
- 270** . Menzel, S., J. Jiang, N. Silver, J. Gallagher, J. Cunningham, G. Surdulescu, . . . S. L. Thein (2007). "The HBS1L-MYB intergenic region on chromosome 6q23.3 influences erythrocyte, platelet, and monocyte counts in humans." Blood **110**(10): 3624-3626.
- 271** . Moayyeri, A., C. J. Hammond, D. J. Hart and T. D. Spector (2012). "The UK Adult Twin Registry (TwinsUK Resource)." Twin Res Hum Genet: 1-6.
- 272** . Moltke, I., N. Grarup, M. E. Jorgensen, P. Bjerregaard, J. T. Treebak, M. Fumagalli, . . . T. Hansen (2014). "A common Greenlandic TBC1D4 variant confers muscle insulin resistance and type 2 diabetes." Nature **512**(7513): 190-193.
- 273** . Monda, K. L., G. K. Chen, K. C. Taylor, C. Palmer, T. L. Edwards, L. A. Lange, . . . C. A. Haiman (2013). "A meta-analysis identifies new loci associated with body mass index in individuals of African ancestry." Nat Genet **45**(6): 690-696.
- 274** . Monteferrario, D., N. A. Bolar, A. E. Marneth, K. M. Hebeda, S. M. Bergevoet, H. Veenstra, . . . B. A. Van der Reijden (2014). "A dominant-negative GFI1B mutation in the gray platelet syndrome." N Engl J Med **370**(3): 245-253.

- 275** . Morgenthaler, S. and W. 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): 28-56.
- 276** . Morrison, A. C., A. Voorman, A. D. Johnson, X. Liu, J. Yu, A. Li, . . . C. Aging Research in Genetic Epidemiology (2013). "Whole-genome sequence-based analysis of high-density lipoprotein cholesterol." *Nat Genet* **45**(8): 899-901.
- 277** . Morton, N. E. (1955). "Sequential tests for the detection of linkage." *Am J Hum Genet* **7**(3): 277-318.
- 278** . Motazacker, M. M., J. Pirruccello, R. Huijgen, R. Do, S. Gabriel, J. Peter, . . . S. W. Fouchier (2012). "Advances in genetics show the need for extending screening strategies for autosomal dominant hypercholesterolaemia." *Eur Heart J* **33**(11): 1360-1366.
- 279** . Musunuru, K., J. P. Pirruccello, R. Do, G. M. Peloso, C. Guiducci, C. Sougnez, . . . S. Kathiresan (2010). "Exome sequencing, ANGPTL3 mutations, and familial combined hypolipidemia." *N Engl J Med* **363**(23): 2220-2227.
- 280** . Musunuru, K., A. Strong, M. Frank-Kamenetsky, N. E. Lee, T. Ahfeldt, K. V. Sachs, . . . D. J. Rader (2010). "From noncoding variant to phenotype via SORT1 at the 1p13 cholesterol locus." *Nature* **466**(7307): 714-719.
- 281** . Nadar, S., A. D. Blann and G. Y. Lip (2004). "Platelet morphology and plasma indices of platelet activation in essential hypertension: effects of amlodipine-based antihypertensive therapy." *Ann Med* **36**(7): 552-557.
- 282** . Naitza, S., E. Porcu, M. Steri, D. D. Taub, A. Mulas, X. Xiao, . . . F. Cucca (2012). "A genome-wide association scan on the levels of markers of inflammation in Sardinians reveals associations that underpin its complex regulation." *PLoS Genet* **8**(1): e1002480.
- 283** . Nalls, M. A., D. J. Couper, T. Tanaka, F. J. van Rooij, M. H. Chen, A. V. Smith, . . . S. K. Ganesh (2011). "Multiple loci are associated with white blood cell phenotypes." *PLoS Genet* **7**(6): e1002113.
- 284** . Navab, M., S. T. Reddy, B. J. Van Lenten and A. M. Fogelman (2011). "HDL and cardiovascular disease: atherogenic and atheroprotective mechanisms." *Nat Rev Cardiol* **8**(4): 222-232.
- 285** . Neale, B. M., M. A. Rivas, B. F. Voight, D. Altshuler, B. Devlin, M. Orho-Melander, . . . M. J. Daly (2011). "Testing for an unusual distribution of rare variants." *PLoS Genet* **7**(3): e1001322.
- 286** . Nicholls, S. J., A. Gordon, J. Johansson, K. Wolski, C. M. Ballantyne, J. J. Kastelein, . . . S. E. Nissen (2011). "Efficacy and safety of a novel oral inducer of apolipoprotein a-I synthesis in statin-treated patients with stable coronary artery disease a randomized controlled trial." *J Am Coll Cardiol* **57**(9): 1111-1119.
- 287** . Nieto, F. J., M. Szklo, A. R. Folsom, R. Rock and M. Mercuri (1992). "Leukocyte count correlates in middle-aged adults: the Atherosclerosis Risk in Communities (ARIC) Study." *Am J Epidemiol* **136**(5): 525-537.
- 288** . Nimptsch, K., K. Aleksandrova, H. Boeing, J. Janke, Y. A. Lee, M. Jenab, . . . T. Pischon (2015). "Association of CRP genetic variants with blood concentrations of C-reactive protein and colorectal cancer risk." *Int J Cancer* **136**(5): 1181-1192.
- 289** . Nordestgaard, B. G., M. Benn, P. Schnohr and A. Tybjaerg-Hansen (2007). "Nonfasting triglycerides and risk of myocardial infarction, ischemic heart disease, and death in men and women." *JAMA* **298**(3): 299-308.
- 290** . Ntalla, I., M. Giannakopoulou, P. Vlachou, K. Giannitsopoulou, V. Gkesou, C. Makridi, . . . G. V. Dedoussis (2014). "Body composition and eating behaviours in relation to dieting involvement in a sample of urban Greek adolescents from the TEENAGE (TEENS of Attica: Genes & Environment) study." *Public Health Nutr* **17**(3): 561-568.
- 291** . Oberdoerffer, S., L. F. Moita, D. Neems, R. P. Freitas, N. Hacohen and A. Rao (2008). "Regulation of CD45 alternative splicing by heterogeneous ribonucleoprotein, hnRNPLL." *Science* **321**(5889): 686-691.

- 292** . Okada, Y., T. Hirota, Y. Kamatani, A. Takahashi, H. Ohmiya, N. Kumasaka, . . . N. Kamatani (2011). "Identification of nine novel loci associated with white blood cell subtypes in a Japanese population." *PLoS Genet* **7**(6): e1002067.
- 293** . Okada, Y., A. Takahashi, H. Ohmiya, N. Kumasaka, Y. Kamatani, N. Hosono, . . . N. Kamatani (2011). "Genome-wide association study for C-reactive protein levels identified pleiotropic associations in the IL6 locus." *Hum Mol Genet* **20**(6): 1224-1231.
- 294** . Olson, R. E. (1998). "Discovery of the lipoproteins, their role in fat transport and their significance as risk factors." *J Nutr* **128**(2 Suppl): 439S-443S.
- 295** . Onengut-Gumuscu, S., W. M. Chen, O. Burren, N. J. Cooper, A. R. Quinlan, J. C. Mychaleckyj, . . . S. S. Rich (2015). "Fine mapping of type 1 diabetes susceptibility loci and evidence for colocalization of causal variants with lymphoid gene enhancers." *Nat Genet* **47**(4): 381-386.
- 296** . Orkin, S. H. and L. I. Zon (2008). "Hematopoiesis: an evolving paradigm for stem cell biology." *Cell* **132**(4): 631-644.
- 297** . Park, J. H., M. H. Gail, C. R. Weinberg, R. J. Carroll, C. C. Chung, Z. Wang, . . . N. Chatterjee (2011). "Distribution of allele frequencies and effect sizes and their interrelationships for common genetic susceptibility variants." *Proc Natl Acad Sci U S A* **108**(44): 18026-18031.
- 298** . Parkes, M., J. C. Barrett, N. J. Prescott, M. Tremelling, C. A. Anderson, S. A. Fisher, . . . C. G. Mathew (2007). "Sequence variants in the autophagy gene IRGM and multiple other replicating loci contribute to Crohn's disease susceptibility." *Nat Genet* **39**(7): 830-832.
- 299** . Pate, R. R., M. Pratt, S. N. Blair, W. L. Haskell, C. A. Macera, C. Bouchard, . . . et al. (1995). "Physical activity and public health. A recommendation from the Centers for Disease Control and Prevention and the American College of Sports Medicine." *JAMA* **273**(5): 402-407.
- 300** . Pathansali, R., N. Smith and P. Bath (2001). "Altered megakaryocyte-platelet haemostatic axis in hypercholesterolaemia." *Platelets* **12**(5): 292-297.
- 301** . Pearson, T. A., G. A. Mensah, R. W. Alexander, J. L. Anderson, R. O. Cannon, 3rd, M. Criqui, . . . A. American Heart (2003). "Markers of inflammation and cardiovascular disease: application to clinical and public health practice: A statement for healthcare professionals from the Centers for Disease Control and Prevention and the American Heart Association." *Circulation* **107**(3): 499-511.
- 302** . Peloso, G. M., P. L. Auer, J. C. Bis, A. Voorman, A. C. Morrison, N. O. Stitzel, . . . L. A. Cupples (2014). "Association of low-frequency and rare coding-sequence variants with blood lipids and coronary heart disease in 56,000 whites and blacks." *Am J Hum Genet* **94**(2): 223-232.
- 303** . Peltola, V., J. Mertsola and O. Ruuskanen (2006). "Comparison of total white blood cell count and serum C-reactive protein levels in confirmed bacterial and viral infections." *J Pediatr* **149**(5): 721-724.
- 304** . Pennisi, E. (2012). "Genomics. ENCODE project writes eulogy for junk DNA." *Science* **337**(6099): 1159, 1161.
- 305** . Pepys, M. B. and G. M. Hirschfield (2003). "C-reactive protein: a critical update." *J Clin Invest* **111**(12): 1805-1812.
- 306** . Persson, M., B. Hedblad, J. J. Nelson and G. Berglund (2007). "Elevated Lp-PLA2 levels add prognostic information to the metabolic syndrome on incidence of cardiovascular events among middle-aged nondiabetic subjects." *Arterioscler Thromb Vasc Biol* **27**(6): 1411-1416.
- 307** . Peterfy, M., O. Ben-Zeev, H. Z. Mao, D. Weissglas-Volkov, B. E. Aouizerat, C. R. Pullinger, . . . M. H. Doolittle (2007). "Mutations in LMF1 cause combined lipase deficiency and severe hypertriglyceridemia." *Nat Genet* **39**(12): 1483-1487.
- 308** . Pickrell, J. K. (2014). "Joint analysis of functional genomic data and genome-wide association studies of 18 human traits." *Am J Hum Genet* **94**(4): 559-573.
- 309** . Pilia, G., W. M. Chen, A. Scuteri, M. Orru, G. Albai, M. Dei, . . . D. Schlessinger (2006). "Heritability of cardiovascular and personality traits in 6,148 Sardinians." *PLoS Genet* **2**(8): e132.
- 310** . Pistis, G., S. U. Okonkwo, M. Traglia, C. Sala, S. Y. Shin, C. Masciullo, . . . D. Toniolo (2013). "Genome wide association analysis of a founder population identified TAF3 as a gene for MCHC in humans." *PLoS One* **8**(7): e69206.

- 311** . Pizzulli, L., A. Yang, J. F. Martin and B. Luderitz (1998). "Changes in platelet size and count in unstable angina compared to stable angina or non-cardiac chest pain." *Eur Heart J* **19**(1): 80-84.
- 312** . Pollin, T. I., C. M. Damcott, H. Shen, S. H. Ott, J. Shelton, R. B. Horenstein, . . . A. R. Shuldiner (2008). "A null mutation in human APOC3 confers a favorable plasma lipid profile and apparent cardioprotection." *Science* **322**(5908): 1702-1705.
- 313** . Power, C. and J. Elliott (2006). "Cohort profile: 1958 British birth cohort (National Child Development Study)." *Int J Epidemiol* **35**(1): 34-41.
- 314** . Price, A. L., G. V. Kryukov, P. I. de Bakker, S. M. Purcell, J. Staples, L. J. Wei and S. R. Sunyaev (2010). "Pooled association tests for rare variants in exon-resequencing studies." *Am J Hum Genet* **86**(6): 832-838.
- 315** . Pritchard, J. K. (2001). "Are rare variants responsible for susceptibility to complex diseases?" *Am J Hum Genet* **69**(1): 124-137.
- 316** . Pritchard, J. K. and N. J. Cox (2002). "The allelic architecture of human disease genes: common disease-common variant...or not?" *Hum Mol Genet* **11**(20): 2417-2423.
- 317** . Prospective Studies, C., S. Lewington, G. Whitlock, R. Clarke, P. Sherliker, J. Emberson, . . . R. Collins (2007). "Blood cholesterol and vascular mortality by age, sex, and blood pressure: a meta-analysis of individual data from 61 prospective studies with 55,000 vascular deaths." *Lancet* **370**(9602): 1829-1839.
- 318** . Qayyum, R., B. M. Snively, E. Ziv, M. A. Nalls, Y. Liu, W. Tang, . . . A. P. Reiner (2012). "A meta-analysis and genome-wide association study of platelet count and mean platelet volume in african americans." *PLoS Genet* **8**(3): e1002491.
- 319** . Quail, M. A., M. Smith, P. Coupland, T. D. Otto, S. R. Harris, T. R. Connor, . . . Y. Gu (2012). "A tale of three next generation sequencing platforms: comparison of Ion Torrent, Pacific Biosciences and Illumina MiSeq sequencers." *BMC Genomics* **13**: 341.
- 320** . Ramanan, V. K., S. L. Risacher, K. Nho, S. Kim, S. Swaminathan, L. Shen, . . . I. Alzheimer's Disease Neuroimaging (2014). "APOE and BCHE as modulators of cerebral amyloid deposition: a florbetapir PET genome-wide association study." *Mol Psychiatry* **19**(3): 351-357.
- 321** . Ramasamy, I. (2014). "Recent advances in physiological lipoprotein metabolism." *Clin Chem Lab Med* **52**(12): 1695-1727.
- 322** . Rana, J. S., B. J. Arsenaault, J. P. Despres, M. Cote, P. J. Talmud, E. Ninio, . . . S. M. Boekholdt (2011). "Inflammatory biomarkers, physical activity, waist circumference, and risk of future coronary heart disease in healthy men and women." *Eur Heart J* **32**(3): 336-344.
- 323** . Rana, J. S., M. Cote, J. P. Despres, M. S. Sandhu, P. J. Talmud, E. Ninio, . . . S. M. Boekholdt (2009). "Inflammatory biomarkers and the prediction of coronary events among people at intermediate risk: the EPIC-Norfolk prospective population study." *Heart* **95**(20): 1682-1687.
- 324** . Rasmussen-Torvik, L. J., J. A. Pacheco, R. A. Wilke, W. K. Thompson, M. D. Ritchie, A. N. Kho, . . . R. L. Chisholm (2012). "High density GWAS for LDL cholesterol in African Americans using electronic medical records reveals a strong protective variant in APOE." *Clin Transl Sci* **5**(5): 394-399.
- 325** . Reich, D., M. A. Nalls, W. H. Kao, E. L. Akylbekova, A. Tandon, N. Patterson, . . . J. G. Wilson (2009). "Reduced neutrophil count in people of African descent is due to a regulatory variant in the Duffy antigen receptor for chemokines gene." *PLoS Genet* **5**(1): e1000360.
- 326** . Reich, D. E. and E. S. Lander (2001). "On the allelic spectrum of human disease." *Trends Genet* **17**(9): 502-510.
- 327** . Reiner, A. P., M. J. Barber, Y. Guan, P. M. Ridker, L. A. Lange, D. I. Chasman, . . . R. M. Krauss (2008). "Polymorphisms of the HNF1A gene encoding hepatocyte nuclear factor-1 alpha are associated with C-reactive protein." *Am J Hum Genet* **82**(5): 1193-1201.
- 328** . Reiner, A. P., S. Beleza, N. Franceschini, P. L. Auer, J. G. Robinson, C. Kooperberg, . . . H. Tang (2012). "Genome-wide association and population genetic analysis of C-reactive protein in African American and Hispanic American women." *Am J Hum Genet* **91**(3): 502-512.

- 329** . Reiner, A. P., G. Lettre, M. A. Nalls, S. K. Ganesh, R. Mathias, M. A. Austin, . . . J. G. Wilson (2011). "Genome-wide association study of white blood cell count in 16,388 African Americans: the continental origins and genetic epidemiology network (COGENT)." *PLoS Genet* **7**(6): e1002108.
- 330** . Ridker, P. M., J. E. Buring and N. Rifai (2001). "Soluble P-selectin and the risk of future cardiovascular events." *Circulation* **103**(4): 491-495.
- 331** . Ridker, P. M., J. E. Buring, N. Rifai and N. R. Cook (2007). "Development and validation of improved algorithms for the assessment of global cardiovascular risk in women: the Reynolds Risk Score." *JAMA* **297**(6): 611-619.
- 332** . Ridker, P. M., M. Cushman, M. J. Stampfer, R. P. Tracy and C. H. Hennekens (1997). "Inflammation, aspirin, and the risk of cardiovascular disease in apparently healthy men." *N Engl J Med* **336**(14): 973-979.
- 333** . Ridker, P. M., E. Danielson, F. A. Fonseca, J. Genest, A. M. Gotto, Jr., J. J. Kastelein, . . . J. S. Group (2008). "Rosuvastatin to prevent vascular events in men and women with elevated C-reactive protein." *N Engl J Med* **359**(21): 2195-2207.
- 334** . Ridker, P. M., E. Danielson, F. A. Fonseca, J. Genest, A. M. Gotto, Jr., J. J. Kastelein, . . . J. T. S. Group (2009). "Reduction in C-reactive protein and LDL cholesterol and cardiovascular event rates after initiation of rosuvastatin: a prospective study of the JUPITER trial." *Lancet* **373**(9670): 1175-1182.
- 335** . Ridker, P. M., G. Pare, A. Parker, R. Y. Zee, J. S. Danik, J. E. Buring, . . . D. I. Chasman (2008). "Loci related to metabolic-syndrome pathways including LEPR, HNF1A, IL6R, and GCKR associate with plasma C-reactive protein: the Women's Genome Health Study." *Am J Hum Genet* **82**(5): 1185-1192.
- 336** . Ridker, P. M., G. Pare, A. N. Parker, R. Y. Zee, J. P. Miletich and D. I. Chasman (2009). "Polymorphism in the CETP gene region, HDL cholesterol, and risk of future myocardial infarction: Genomewide analysis among 18 245 initially healthy women from the Women's Genome Health Study." *Circ Cardiovasc Genet* **2**(1): 26-33.
- 337** . Ridker, P. M., N. P. Paynter, N. Rifai, J. M. Gaziano and N. R. Cook (2008). "C-reactive protein and parental history improve global cardiovascular risk prediction: the Reynolds Risk Score for men." *Circulation* **118**(22): 2243-2251, 2244p following 2251.
- 338** . Ridker, P. M., N. Rifai, M. Clearfield, J. R. Downs, S. E. Weis, J. S. Miles, . . . I. Air Force/Texas Coronary Atherosclerosis Prevention Study (2001). "Measurement of C-reactive protein for the targeting of statin therapy in the primary prevention of acute coronary events." *N Engl J Med* **344**(26): 1959-1965.
- 339** . Ridker, P. M., N. Rifai, M. J. Stampfer and C. H. Hennekens (2000). "Plasma concentration of interleukin-6 and the risk of future myocardial infarction among apparently healthy men." *Circulation* **101**(15): 1767-1772.
- 340** . Rimm, E. B., E. L. Giovannucci, W. C. Willett, G. A. Colditz, A. Ascherio, B. Rosner and M. J. Stampfer (1991). "Prospective study of alcohol consumption and risk of coronary disease in men." *Lancet* **338**(8765): 464-468.
- 341** . Rios, J., E. Stein, J. Shendure, H. H. Hobbs and J. C. Cohen (2010). "Identification by whole-genome resequencing of gene defect responsible for severe hypercholesterolemia." *Hum Mol Genet* **19**(22): 4313-4318.
- 342** . Risch, N. and K. Merikangas (1996). "The future of genetic studies of complex human diseases." *Science* **273**(5281): 1516-1517.
- 343** . Robinson, J. G. (2009). "Are you targeting non-high-density lipoprotein cholesterol?" *J Am Coll Cardiol* **55**(1): 42-44.
- 344** . Robinson, M. R., N. R. Wray and P. M. Visscher (2014). "Explaining additional genetic variation in complex traits." *Trends Genet* **30**(4): 124-132.
- 345** . Rosenthal, E. A., J. Ranchalis, D. R. Crosslin, A. Burt, J. D. Brunzell, A. G. Motulsky, . . . G. P. Jarvik (2013). "Joint linkage and association analysis with exome sequence data implicates SLC25A40 in hypertriglyceridemia." *Am J Hum Genet* **93**(6): 1035-1045.

- 346** . Ruchat, S. M., J. P. Despres, S. J. Weisnagel, Y. C. Chagnon, C. Bouchard and L. Perusse (2008). "Genome-wide linkage analysis for circulating levels of adipokines and C-reactive protein in the Quebec family study (QFS)." J Hum Genet **53**(7): 629-636.
- 347** . Ruggiero, C., E. J. Metter, A. Cherubini, M. Maggio, R. Sen, S. S. Najjar, . . . L. Ferrucci (2007). "White blood cell count and mortality in the Baltimore Longitudinal Study of Aging." J Am Coll Cardiol **49**(18): 1841-1850.
- 348** . Rust, S., M. Rosier, H. Funke, J. Real, Z. Amoura, J. C. Piette, . . . G. Assmann (1999). "Tangier disease is caused by mutations in the gene encoding ATP-binding cassette transporter 1." Nat Genet **22**(4): 352-355.
- 349** . Sabatti, C., S. K. Service, A. L. Hartikainen, A. Pouta, S. Ripatti, J. Brodsky, . . . L. Peltonen (2009). "Genome-wide association analysis of metabolic traits in a birth cohort from a founder population." Nat Genet **41**(1): 35-46.
- 350** . Sandhu, M. S., D. M. Waterworth, S. L. Debenham, E. Wheeler, K. Papadakis, J. H. Zhao, . . . V. Mooser (2008). "LDL-cholesterol concentrations: a genome-wide association study." Lancet **371**(9611): 483-491.
- 351** . Sankaran, V. G., L. S. Ludwig, E. Sicinska, J. Xu, D. E. Bauer, J. C. Eng, . . . H. F. Lodish (2012). "Cyclin D3 coordinates the cell cycle during differentiation to regulate erythrocyte size and number." Genes Dev **26**(18): 2075-2087.
- 352** . Sankaran, V. G., T. F. Menne, J. Xu, T. E. Akie, G. Lettre, B. Van Handel, . . . S. H. Orkin (2008). "Human fetal hemoglobin expression is regulated by the developmental stage-specific repressor BCL11A." Science **322**(5909): 1839-1842.
- 353** . Sankaran, V. G., J. Xu and S. H. Orkin (2010). "Advances in the understanding of haemoglobin switching." Br J Haematol **149**(2): 181-194.
- 354** . Santos-Cortez, R. L., K. Lee, A. P. Giese, M. Ansar, M. Amin-Ud-Din, K. Rehn, . . . S. M. Leal (2014). "Adenylate cyclase 1 (ADCY1) mutations cause recessive hearing impairment in humans and defects in hair cell function and hearing in zebrafish." Hum Mol Genet **23**(12): 3289-3298.
- 355** . Santos, S., T. W. Rooke, K. R. Bailey, J. P. McConnell and I. J. Kullo (2004). "Relation of markers of inflammation (C-reactive protein, white blood cell count, and lipoprotein-associated phospholipase A2) to the ankle-brachial index." Vasc Med **9**(3): 171-176.
- 356** . Sarwar, N., J. Danesh, G. Eiriksdottir, G. Sigurdsson, N. Wareham, S. Bingham, . . . V. Gudnason (2007). "Triglycerides and the risk of coronary heart disease: 10,158 incident cases among 262,525 participants in 29 Western prospective studies." Circulation **115**(4): 450-458.
- 357** . Sattar, N., H. M. Murray, A. McConnachie, G. J. Blauw, E. L. Bollen, B. M. Buckley, . . . P. S. Group (2007). "C-reactive protein and prediction of coronary heart disease and global vascular events in the Prospective Study of Pravastatin in the Elderly at Risk (PROSPER)." Circulation **115**(8): 981-989.
- 358** . Saxena, R., B. F. Voight, V. Lyssenko, N. P. Burtt, P. I. de Bakker, H. Chen, . . . S. Purcell (2007). "Genome-wide association analysis identifies loci for type 2 diabetes and triglyceride levels." Science **316**(5829): 1331-1336.
- 359** . Scheet, P. and M. Stephens (2006). "A fast and flexible statistical model for large-scale population genotype data: applications to inferring missing genotypes and haplotypic phase." Am J Hum Genet **78**(4): 629-644.
- 360** . Schunkert, H., I. R. Konig, S. Kathiresan, M. P. Reilly, T. L. Assimes, H. Holm, . . . N. J. Samani (2011). "Large-scale association analysis identifies 13 new susceptibility loci for coronary artery disease." Nat Genet **43**(4): 333-338.
- 361** . Seddon, J. M., R. Reynolds, J. Maller, J. A. Fagerness, M. J. Daly and B. Rosner (2009). "Prediction model for prevalence and incidence of advanced age-related macular degeneration based on genetic, demographic, and environmental variables." Invest Ophthalmol Vis Sci **50**(5): 2044-2053.

- 362** . Selhub, J., P. F. Jacques, A. G. Bostom, R. B. D'Agostino, P. W. Wilson, A. J. Belanger, . . . I. H. Rosenberg (1995). "Association between plasma homocysteine concentrations and extracranial carotid-artery stenosis." *N Engl J Med* **332**(5): 286-291.
- 363** . Service., U. P. H. (1983). "The Health Consequences of Smoking: Cardiovascular Disease: A Report of the Surgeon General. ." DHHS (PHS) 84-50204.
- 364** . Shameer, K., J. C. Denny, K. Ding, H. Jouni, D. R. Crosslin, M. de Andrade, . . . I. J. Kullo (2014). "A genome- and phenome-wide association study to identify genetic variants influencing platelet count and volume and their pleiotropic effects." *Hum Genet* **133**(1): 95-109.
- 365** . Shankar, A., J. J. Wang, E. Rohtchina, M. C. Yu, R. Kefford and P. Mitchell (2006). "Association between circulating white blood cell count and cancer mortality: a population-based cohort study." *Arch Intern Med* **166**(2): 188-194.
- 366** . Sharp, D., L. Blinderman, K. A. Combs, B. Kienzle, B. Ricci, K. Wager-Smith, . . . et al. (1993). "Cloning and gene defects in microsomal triglyceride transfer protein associated with abetalipoproteinaemia." *Nature* **365**(6441): 65-69.
- 367** . Shepherd, J., S. M. Cobbe, I. Ford, C. G. Isles, A. R. Lorimer, P. W. MacFarlane, . . . C. J. Packard (1995). "Prevention of coronary heart disease with pravastatin in men with hypercholesterolemia. West of Scotland Coronary Prevention Study Group." *N Engl J Med* **333**(20): 1301-1307.
- 368** . Soranzo, N., A. Rendon, C. Gieger, C. I. Jones, N. A. Watkins, S. Menzel, . . . W. H. Ouwehand (2009). "A novel variant on chromosome 7q22.3 associated with mean platelet volume, counts, and function." *Blood* **113**(16): 3831-3837.
- 369** . Soranzo, N., F. Rivadeneira, U. Chinappen-Horsley, I. Malkina, J. B. Richards, N. Hammond, . . . P. Deloukas (2009). "Meta-analysis of genome-wide scans for human adult stature identifies novel Loci and associations with measures of skeletal frame size." *PLoS Genet* **5**(4): e1000445.
- 370** . Soranzo, N., T. D. Spector, M. Mangino, B. Kuhnel, A. Rendon, A. Teumer, . . . C. Gieger (2009). "A genome-wide meta-analysis identifies 22 loci associated with eight hematological parameters in the HaemGen consortium." *Nat Genet* **41**(11): 1182-1190.
- 371** . Soria, L. F., E. H. Ludwig, H. R. Clarke, G. L. Vega, S. M. Grundy and B. J. McCarthy (1989). "Association between a specific apolipoprotein B mutation and familial defective apolipoprotein B-100." *Proc Natl Acad Sci U S A* **86**(2): 587-591.
- 372** . Sorrentino, V., S. W. Fouchier, M. M. Motazacker, J. K. Nelson, J. C. Defesche, G. M. Dallinga-Thie, . . . N. Zelcer (2013). "Identification of a loss-of-function inducible degrader of the low-density lipoprotein receptor variant in individuals with low circulating low-density lipoprotein." *Eur Heart J* **34**(17): 1292-1297.
- 373** . Spector, T. D. and F. M. Williams (2006). "The UK Adult Twin Registry (TwinsUK)." *Twin Res Hum Genet* **9**(6): 899-906.
- 374** . Spencer, C. C., Z. Su, P. Donnelly and J. Marchini (2009). "Designing genome-wide association studies: sample size, power, imputation, and the choice of genotyping chip." *PLoS Genet* **5**(5): e1000477.
- 375** . St George-Hyslop, P. H., R. E. Tanzi, R. J. Polinsky, J. L. Haines, L. Nee, P. C. Watkins, . . . et al. (1987). "The genetic defect causing familial Alzheimer's disease maps on chromosome 21." *Science* **235**(4791): 885-890.
- 376** . Stampfer, M. J., G. A. Colditz, W. C. Willett, F. E. Speizer and C. H. Hennekens (1988). "A prospective study of moderate alcohol consumption and the risk of coronary disease and stroke in women." *N Engl J Med* **319**(5): 267-273.
- 377** . Stein, E. A., S. Mellis, G. D. Yancopoulos, N. Stahl, D. Logan, W. B. Smith, . . . G. D. Swergold (2012). "Effect of a monoclonal antibody to PCSK9 on LDL cholesterol." *N Engl J Med* **366**(12): 1108-1118.
- 378** . Stephens, M. (2013). "A unified framework for association analysis with multiple related phenotypes." *PLoS One* **8**(7): e65245.
- 379** . Stitzel, N. O., S. W. Fouchier, B. Sjouke, G. M. Peloso, A. M. Moscoso, P. L. Auer, . . . G. O. E. S. P. Blood Institute (2013). "Exome sequencing and directed clinical phenotyping diagnose cholesterol

ester storage disease presenting as autosomal recessive hypercholesterolemia." Arterioscler Thromb Vasc Biol **33**(12): 2909-2914.

380 . Stolk, L., J. R. Perry, D. I. Chasman, C. He, M. Mangino, P. Sulem, . . . K. L. Lunetta (2012). "Meta-analyses identify 13 loci associated with age at menopause and highlight DNA repair and immune pathways." Nat Genet **44**(3): 260-268.

381 . Surakka, I., A. Isaacs, L. C. Karssen, P. P. Laurila, R. P. Middelberg, E. Tikkanen, . . . E. Consortium (2011). "A genome-wide screen for interactions reveals a new locus on 4p15 modifying the effect of waist-to-hip ratio on total cholesterol." PLoS Genet **7**(10): e1002333.

382 . Surakka, I., J. B. Whitfield, M. Perola, P. M. Visscher, G. W. Montgomery, M. Falchi, . . . E. P. Genom (2012). "A genome-wide association study of monozygotic twin-pairs suggests a locus related to variability of serum high-density lipoprotein cholesterol." Twin Res Hum Genet **15**(6): 691-699.

383 . Syvanen, A. C. (2005). "Toward genome-wide SNP genotyping." Nat Genet **37** **Suppl**: S5-10.

384 . Szmítko, P. E., C. H. Wang, R. D. Weisel, J. R. de Almeida, T. J. Anderson and S. Verma (2003). "New markers of inflammation and endothelial cell activation: Part I." Circulation **108**(16): 1917-1923.

385 . Tachmazidou, I., G. Dedoussis, L. Southam, A. E. Farmaki, G. R. Ritchie, D. K. Xifara, . . . E. Zeggini (2013). "A rare functional cardioprotective APOC3 variant has risen in frequency in distinct population isolates." Nat Commun **4**: 2872.

386 . Tan, A., J. Sun, N. Xia, X. Qin, Y. Hu, S. Zhang, . . . J. Xu (2012). "A genome-wide association and gene-environment interaction study for serum triglycerides levels in a healthy Chinese male population." Hum Mol Genet **21**(7): 1658-1664.

387 . Tchernitchko, D., M. Goossens and H. Wajcman (2004). "In silico prediction of the deleterious effect of a mutation: proceed with caution in clinical genetics." Clin Chem **50**(11): 1974-1978.

388 . Teo, K. K., S. Ounpuu, S. Hawken, M. R. Pandey, V. Valentin, D. Hunt, . . . I. S. Investigators (2006). "Tobacco use and risk of myocardial infarction in 52 countries in the INTERHEART study: a case-control study." Lancet **368**(9536): 647-658.

389 . Teo, Y. Y., K. S. Small and D. P. Kwiatkowski (2010). "Methodological challenges of genome-wide association analysis in Africa." Nat Rev Genet **11**(2): 149-160.

390 . Teslovich, T. M., K. Musunuru, A. V. Smith, A. C. Edmondson, I. M. Stylianou, M. Koseki, . . . S. Kathiresan (2010). "Biological, clinical and population relevance of 95 loci for blood lipids." Nature **466**(7307): 707-713.

391 . Tg, N. H. L. Hdl Working Group of the Exome Sequencing Project, I. Blood, J. Crosby, G. M. Peloso, P. L. Auer, . . . S. Kathiresan (2014). "Loss-of-function mutations in APOC3, triglycerides, and coronary disease." N Engl J Med **371**(1): 22-31.

392 . Thaulow, E., J. Erikssen, L. Sandvik, H. Stormorken and P. F. Cohn (1991). "Blood platelet count and function are related to total and cardiovascular death in apparently healthy men." Circulation **84**(2): 613-617.

393 . The TG and HDL Working Group of the Exome Sequencing Project, N. H. L. B., Institute (2014). "Loss-of-Function Mutations in APOC3, Triglycerides, and Coronary Disease." N Engl J Med **371**(1): 22-31.

394 . The UK10K Consortium (2015). "The UK10K project: rare variants in health and disease." *submitted*.

395 . The Women's Health Initiative Study Group (1998). "Design of the Women's Health Initiative clinical trial and observational study. The Women's Health Initiative Study Group." Control Clin Trials **19**(1): 61-109.

396 . Thompson, D., M. B. Pepys and S. P. Wood (1999). "The physiological structure of human C-reactive protein and its complex with phosphocholine." Structure **7**(2): 169-177.

397 . Thomson, W., A. Barton, X. Ke, S. Eyre, A. Hinks, J. Bowes, . . . J. Worthington (2007). "Rheumatoid arthritis association at 6q23." Nat Genet **39**(12): 1431-1433.

- 398** . Timmann, C., T. Thye, M. Vens, J. Evans, J. May, C. Ehmen, . . . R. D. Horstmann (2012). "Genome-wide association study indicates two novel resistance loci for severe malaria." Nature **489**(7416): 443-446.
- 399** . Timpson, N., K. Walter, M. JL, I. Tachmazidou, G. Malerba, S.-Y. Shin, . . . N. Soranzo "A novel low-frequency variant near APOC3 is associated with plasma triglyceride and VLDL levels in Europeans." Nature Communications (Under peer review).
- 400** . Timpson, N. J., K. Walter, J. L. Min, I. Tachmazidou, G. Malerba, S. Y. Shin, . . . U. O. C. Members (2014). "A rare variant in APOC3 is associated with plasma triglyceride and VLDL levels in Europeans." Nat Commun **5**: 4871.
- 401** . Tiong, A. Y. and D. Brieger (2005). "Inflammation and coronary artery disease." Am Heart J **150**(1): 11-18.
- 402** . Todd, J. A., N. M. Walker, J. D. Cooper, D. J. Smyth, K. Downes, V. Plagnol, . . . D. G. Clayton (2007). "Robust associations of four new chromosome regions from genome-wide analyses of type 1 diabetes." Nat Genet **39**(7): 857-864.
- 403** . Traglia, M., C. Sala, C. Masciullo, V. Cverhova, F. Lori, G. Pistis, . . . D. Toniolo (2009). "Heritability and Demographic Analyses in the Large Isolated Population of Val Borbera Suggest Advantages in Mapping Complex Traits Genes." PLoS ONE **4**(10): e7554.
- 404** . Triglyceride Coronary Disease Genetics, C., C. Emerging Risk Factors, N. Sarwar, M. S. Sandhu, S. L. Ricketts, A. S. Butterworth, . . . J. Danesh (2010). "Triglyceride-mediated pathways and coronary disease: collaborative analysis of 101 studies." Lancet **375**(9726): 1634-1639.
- 405** . Tunstall-Pedoe, H., M. Woodward and S. g. o. r. estimation (2006). "By neglecting deprivation, cardiovascular risk scoring will exacerbate social gradients in disease." Heart **92**(3): 307-310.
- 406** . Uda, M., R. Galanello, S. Sanna, G. Lettre, V. G. Sankaran, W. Chen, . . . A. Cao (2008). "Genome-wide association study shows BCL11A associated with persistent fetal hemoglobin and amelioration of the phenotype of beta-thalassemia." Proc Natl Acad Sci U S A **105**(5): 1620-1625.
- 407** . van der Harst, P., W. Zhang, I. Mateo Leach, A. Rendon, N. Verweij, J. Sehmi, . . . J. C. Chambers (2012). "Seventy-five genetic loci influencing the human red blood cell." Nature **492**(7429): 369-375.
- 408** . van Dongen, J., G. Willemsen, W. M. Chen, E. J. de Geus and D. I. Boomsma (2013). "Heritability of metabolic syndrome traits in a large population-based sample." J Lipid Res **54**(10): 2914-2923.
- 409** . Venter, J. C., M. D. Adams, E. W. Myers, P. W. Li, R. J. Mural, G. G. Sutton, . . . X. Zhu (2001). "The sequence of the human genome." Science **291**(5507): 1304-1351.
- 410** . Vinayagamoorthy, N., H. J. Hu, S. H. Yim, S. H. Jung, J. Jo, S. H. Jee and Y. J. Chung (2014). "New variants including ARG1 polymorphisms associated with C-reactive protein levels identified by genome-wide association and pathway analysis." PLoS One **9**(4): e95866.
- 411** . Visscher, P. M., M. A. Brown, M. I. McCarthy and J. Yang (2012). "Five years of GWAS discovery." Am J Hum Genet **90**(1): 7-24.
- 412** . Visscher, P. M., M. E. Goddard, E. M. Derks and N. R. Wray (2012). "Evidence-based psychiatric genetics, AKA the false dichotomy between common and rare variant hypotheses." Mol Psychiatry **17**(5): 474-485.
- 413** . Voight, B. F., G. M. Peloso, M. Orho-Melander, R. Frikke-Schmidt, M. Barbalic, M. K. Jensen, . . . S. Kathiresan (2012). "Plasma HDL cholesterol and risk of myocardial infarction: a mendelian randomisation study." Lancet **380**(9841): 572-580.
- 414** . von Eckardstein, A., H. Funke, A. Henke, K. Altland, A. Benninghoven and G. Assmann (1989). "Apolipoprotein A-I variants. Naturally occurring substitutions of proline residues affect plasma concentration of apolipoprotein A-I." J Clin Invest **84**(6): 1722-1730.
- 415** . Wallace, C., S. J. Newhouse, P. Braund, F. Zhang, M. Tobin, M. Falchi, . . . P. B. Munroe (2008). "Genome-wide association study identifies genes for biomarkers of cardiovascular disease: serum urate and dyslipidemia." Am J Hum Genet **82**(1): 139-149.
- 416** . Waterworth, D. M., S. L. Ricketts, K. Song, L. Chen, J. H. Zhao, S. Ripatti, . . . M. S. Sandhu (2010). "Genetic variants influencing circulating lipid levels and risk of coronary artery disease." Arterioscler Thromb Vasc Biol **30**(11): 2264-2276.

- 417** . Watowich, S. S., X. Xie, U. Klingmuller, J. Kere, M. Lindlof, S. Berglund and A. de la Chapelle (1999). "Erythropoietin receptor mutations associated with familial erythrocytosis cause hypersensitivity to erythropoietin in the heterozygous state." Blood **94**(7): 2530-2532.
- 418** . Webb, J., H. Gonna and K. K. Ray (2013). "Lipid management: maximising reduction of cardiac risk." Clin Med **13**(6): 618-620.
- 419** . Weiss, L. A., L. Pan, M. Abney and C. Ober (2006). "The sex-specific genetic architecture of quantitative traits in humans." Nat Genet **38**(2): 218-222.
- 420** . Weissglas-Volkov, D., C. A. Aguilar-Salinas, E. Nikkola, K. A. Deere, I. Cruz-Bautista, O. Arellano-Campos, . . . P. Pajukanta (2013). "Genomic study in Mexicans identifies a new locus for triglycerides and refines European lipid loci." J Med Genet **50**(5): 298-308.
- 421** . Wellcome Trust Case Control, C. (2007). "Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls." Nature **447**(7145): 661-678.
- 422** . Wellcome Trust Case Control, C., N. Craddock, M. E. Hurles, N. Cardin, R. D. Pearson, V. Plagnol, . . . P. Donnelly (2010). "Genome-wide association study of CNVs in 16,000 cases of eight common diseases and 3,000 shared controls." Nature **464**(7289): 713-720.
- 423** . Wellcome Trust Case Control, C., J. B. Maller, G. McVean, J. Byrnes, D. Vukcevic, K. Palin, . . . P. Donnelly (2012). "Bayesian refinement of association signals for 14 loci in 3 common diseases." Nat Genet **44**(12): 1294-1301.
- 424** . 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): 661-678.
- 425** . Westra, H. J., M. J. Peters, T. Esko, H. Yaghootkar, C. Schurmann, J. Kettunen, . . . L. Franke (2013). "Systematic identification of trans eQTLs as putative drivers of known disease associations." Nat Genet **45**(10): 1238-1243.
- 426** . Whittall, R. A., S. Matheus, T. Cranston, G. J. Miller and S. E. Humphries (2002). "The intron 14 2140+5G>A variant in the low density lipoprotein receptor gene has no effect on plasma cholesterol levels." J Med Genet **39**(9): e57.
- 427** . Willems, J. M., S. Trompet, G. J. Blauw, R. G. Westendorp and A. J. de Craen (2010). "White blood cell count and C-reactive protein are independent predictors of mortality in the oldest old." J Gerontol A Biol Sci Med Sci **65**(7): 764-768.
- 428** . Willer, C. J., Y. Li and G. R. Abecasis (2010). "METAL: fast and efficient meta-analysis of genomewide association scans." Bioinformatics **26**(17): 2190-2191.
- 429** . Willer, C. J., S. Sanna, A. U. Jackson, A. Scuteri, L. L. Bonnycastle, R. Clarke, . . . G. R. Abecasis (2008). "Newly identified loci that influence lipid concentrations and risk of coronary artery disease." Nat Genet **40**(2): 161-169.
- 430** . Willer, C. J., E. M. Schmidt, S. Sengupta, G. M. Peloso, S. Gustafsson, S. Kanoni, . . . G. R. Abecasis (2013). "Discovery and refinement of loci associated with lipid levels." Nat Genet **45**(11): 1274-1283.
- 431** . Wilson, P. W., R. B. D'Agostino, D. Levy, A. M. Belanger, H. Silbershatz and W. B. Kannel (1998). "Prediction of coronary heart disease using risk factor categories." Circulation **97**(18): 1837-1847.
- 432** . Wilson, P. W., B. H. Nam, M. Pencina, R. B. D'Agostino, Sr., E. J. Benjamin and C. J. O'Donnell (2005). "C-reactive protein and risk of cardiovascular disease in men and women from the Framingham Heart Study." Arch Intern Med **165**(21): 2473-2478.
- 433** . Winkelmann, B. R., W. Marz, B. O. Boehm, R. Zotz, J. Hager, P. Hellstern, . . . L. S. Group (2001). "Rationale and design of the LURIC study--a resource for functional genomics, pharmacogenomics and long-term prognosis of cardiovascular disease." Pharmacogenomics **2**(1 Suppl 1): S1-73.
- 434** . Wolfs, M. G., M. H. Hofker, C. Wijmenga and T. W. van Haeften (2009). "Type 2 Diabetes Mellitus: New Genetic Insights will Lead to New Therapeutics." Curr Genomics **10**(2): 110-118.
- 435** . Wong, N. D. (2014). "Epidemiological studies of CHD and the evolution of preventive cardiology." Nat Rev Cardiol **11**(5): 276-289.

- 436** . Wood, A. R., T. Esko, J. Yang, S. Vedantam, T. H. Pers, S. Gustafsson, . . . T. M. Frayling (2014). "Defining the role of common variation in the genomic and biological architecture of adult human height." Nat Genet **46**(11): 1173-1186.
- 437** . Wu, M. C., S. Lee, T. Cai, Y. Li, M. Boehnke and X. Lin (2011). "Rare-variant association testing for sequencing data with the sequence kernel association test." American journal of human genetics **89**(1): 82-93.
- 438** . Wu, M. C., S. Lee, T. Cai, Y. Li, M. Boehnke and X. Lin (2011). "Rare-variant association testing for sequencing data with the sequence kernel association test." Am J Hum Genet **89**(1): 82-93.
- 439** . Wu, Y., A. F. Marvelle, J. Li, D. C. Croteau-Chonka, A. B. Feranil, C. W. Kuzawa, . . . K. L. Mohlke (2013). "Genetic association with lipids in Filipinos: waist circumference modifies an APOA5 effect on triglyceride levels." J Lipid Res **54**(11): 3198-3205.
- 440** . Wu, Y., T. W. McDade, C. W. Kuzawa, J. Borja, Y. Li, L. S. Adair, . . . L. A. Lange (2012). "Genome-wide association with C-reactive protein levels in CLHNS: evidence for the CRP and HNF1A loci and their interaction with exposure to a pathogenic environment." Inflammation **35**(2): 574-583.
- 441** . Xu, C., I. Tachmazidou, K. Walter, A. Ciampi, E. Zeggini, C. M. T. Greenwood and t. U. K. Consortium (2014). "Estimating Genome-Wide Significance for Whole-Genome Sequencing Studies." Genetic Epidemiology: n/a-n/a.
- 442** . Xu, J., C. Peng, V. G. Sankaran, Z. Shao, E. B. Esrick, B. G. Chong, . . . S. H. Orkin (2011). "Correction of sickle cell disease in adult mice by interference with fetal hemoglobin silencing." Science **334**(6058): 993-996.
- 443** . Yan, J., T. Takahashi, T. Ohura, H. Adachi, I. Takahashi, E. Ogawa, . . . A. Koizumi (2013). "Combined linkage analysis and exome sequencing identifies novel genes for familial goiter." J Hum Genet **58**(6): 366-377.
- 444** . Yang, J., B. Benyamin, B. P. McEvoy, S. Gordon, A. K. Henders, D. R. Nyholt, . . . P. M. Visscher (2010). "Common SNPs explain a large proportion of the heritability for human height." Nat Genet **42**(7): 565-569.
- 445** . Yang, J., T. A. Manolio, L. R. Pasquale, E. Boerwinkle, N. Caporaso, J. M. Cunningham, . . . P. M. Visscher (2011). "Genome partitioning of genetic variation for complex traits using common SNPs." Nat Genet **43**(6): 519-525.
- 446** . Yang, Q., S. Kathiresan, J. P. Lin, G. H. Tofler and C. J. O'Donnell (2007). "Genome-wide association and linkage analyses of hemostatic factors and hematological phenotypes in the Framingham Heart Study." BMC Med Genet **8 Suppl 1**: S12.
- 447** . Young, S. G., S. J. Bertics, L. K. Curtiss, B. W. Dubois and J. L. Witztum (1987). "Genetic analysis of a kindred with familial hypobetalipoproteinemia. Evidence for two separate gene defects: one associated with an abnormal apolipoprotein B species, apolipoprotein B-37; and a second associated with low plasma concentrations of apolipoprotein B-100." J Clin Invest **79**(6): 1842-1851.
- 448** . Yusuf, S., S. Hawken, S. Ounpuu, T. Dans, A. Avezum, F. Lanas, . . . I. S. Investigators (2004). "Effect of potentially modifiable risk factors associated with myocardial infarction in 52 countries (the INTERHEART study): case-control study." Lancet **364**(9438): 937-952.
- 449** . Zeggini, E. (2014). "Genetic characterisation of Greek population isolates reveals strong genetic drift at missense and trait-associated variants." under review.
- 450** . Zeggini, E. (2014). "Using genetically isolated populations to understand the genomic basis of disease." Genome Med **6**(10): 83.
- 451** . Zeggini, E., M. N. Weedon, C. M. Lindgren, T. M. Frayling, K. S. Elliott, H. Lango, . . . A. T. Hattersley (2007). "Replication of genome-wide association signals in UK samples reveals risk loci for type 2 diabetes." Science **316**(5829): 1336-1341.
- 452** . Zelcer, N., C. Hong, R. Boyadjian and P. Tontonoz (2009). "LXR regulates cholesterol uptake through Idol-dependent ubiquitination of the LDL receptor." Science **325**(5936): 100-104.
- 453** . Zeng, S. M., J. Yankowitz, J. A. Widness and R. G. Strauss (2001). "Etiology of differences in hematocrit between males and females: sequence-based polymorphisms in erythropoietin and its receptor." J Genet Specif Med **4**(1): 35-40.

- 454** . Zhou, L., M. He, Z. Mo, C. Wu, H. Yang, D. Yu, . . . T. Wu (2013). "A genome wide association study identifies common variants associated with lipid levels in the Chinese population." PLoS One **8**(12): e82420.
- 455** . Zhou, X. and M. Stephens (2012). "Genome-wide efficient mixed-model analysis for association studies." Nat Genet **44**(7): 821-824.
- 456** . Zuk, O., S. F. Schaffner, K. Samocha, R. Do, E. Hechter, S. Kathiresan, . . . E. S. Lander (2014). "Searching for missing heritability: designing rare variant association studies." Proc Natl Acad Sci U S A **111**(4): E455-464.
- 457** . Zwaka, T. P., V. Hombach and J. Torzewski (2001). "C-reactive protein-mediated low density lipoprotein uptake by macrophages: implications for atherosclerosis." Circulation **103**(9): 1194-1197.

