A Bibliographic Introduction to the Common Diseases Program of the Institute for Translational Genomics and Population Sciences Lundquist Institute (formerly LABiomed) at Harbor-UCLA Medical Center

December 7, 2020

The following is a bibliographic introduction to the Genomics of Common Disease Program of the Institute for Translational Genomics and Population Sciences, located at the Lundquist Institute for Biomedical Innovation (formerly Los Angeles Biomedical Research Institute) at Harbor-UCLA Medical Center (and academically located in the Departments of Pediatrics and Medicine at Harbor-UCLA Medical Center). The Genomics Institute was founded in June/July of 2013, and therefore has been in existence slightly more than 6 and a half years.

There are 6 faculty in the Common Disease Genetics Program – Professor Yii-Der Ida Chen, Professor Xiuqing Guo, Associate Professor Xiaohui Li, Professor Henry Lin, Professor Kent D. Taylor, and Professor Jerome I. Rotter.

Their research is in the genetics of common, complex diseases, i.e. the earliest determinants of what are for the most part adult diseases but with the origins of their pathophysiology in childhood. They have and are contributing to our knowledge of the genetic basis of cardiometabolic disorders (atherosclerosis, coronary artery disease, valvular heart disease, arrhythmias and EKG variation, blood pressure and hypertension, lipid disorders, nonalcoholic fatty liver disease, obesity, diabetes, diabetic kidney disease, diabetic eye disease, and insulin resistance), eye diseases (diabetic retinopathy, keratoconus, glaucoma, macular degeneration, retinopathy of prematurity, retinal vasculature), and pharmacogenetics (genetic determinants of response to a therapy). Their work (which is intensively collaborative) has utilized a variety of paradigms, from family based, to case-control, to cohort, to pharmacogenetic studies, and from candidate gene, to family based linkage, to genome-wide association, to large scale specialized genotyping and sequencing, to whole exome and whole genome sequencing. These studies have included deep phenotyping and biochemical analyses, and have now entered the multi-omics era with studies of methylomics, transcriptomics, metabolomics, and proteomics. These studies involve hundreds to hundreds of thousands of subjects and from a handful to hundreds of investigators, the epitome of what is termed team science. Members of the Genomics Institute have been especially active in multiethnic studies including those in Caucasian, Hispanic, African-Americans and Chinese populations. In the process, they helped delineate the genetic architecture of diabetes and insulin resistance, of blood pressure and hypertension, of lipid disorders and coronary artery disease, of cardiac arrhythmias and EKG variation and suddent cardiac death, of obesity and fatty liver, and of diabetic retinopathy and glaucoma, in multiple ethnic groups. The ultimate goal of this work is to identify the optimal therapy and prevention for cardiometabolic and ocular disorders as a function of an individual's genetic predispositions. Thus, this is the basis for precision/personalized medicine, especially in minority populations.

Since the beginning of the program at Lundquist/Harbor-UCLA in mid-2013, the 6 faculty have contributed to some 526 peer review publications (for the entire 10 faculty of the Genomics Institute, the total is 580). To introduce the program, we have listed, as examples, 100 of these publications, 55 of which have been published in the world's leading biomedical journals, i.e. the New England Journal of Medicine, Journal of the American Medical Association, Nature, Science, Cell, and Nature Genetics. An additional 28 have been published in journals with an impact factor greater than 10 and often the lead specialty journal in its field (American Journal of Human Genetics, Cell Metabolism, Circulation, Circulation Research, Diabetes Care, European Heart Journal, Genome Research, Journal of Clinical Investigation, Journal of the American College of Cardiology, Nature Communications, PLOS Medicine, and Science Translational Medicine). (Any individual article can be obtained by inquiring to Daune Thorington, daunet@lundquist.org, as well as the list of the entire 526 or 580 papers respectively.) These papers are listed in 10 scientific areas related to diabetes (and associated obesity), cardiovascular disease (including EKG variation), as well as its risk factors (lipids, hypertension), ocular disorders, and genetics of response to therapeutics for these disorders (the discipline of pharmacogenetics), as well as selected other common diseases and population genetics papers.

Publications Table of Contents (by subject)

A. **Diabetes Mellitus** (Includes Type 2DM, Insulin Resistance, Insulin Secretion, Insulin Clearance, Fatty Liver Disease, Diabetic Nephropathy) (18 examples)

1. Kuo, J.Z., Sheu, W.H.-H., Assimes, T.L… Wang, T.-D., Guo, X., Taylor, K.D., Chuang, L.-M… Quertermous, T., Rotter, J.I., Chen, Y.-D.I.: *Trans-ethnic fine mapping identifies a novel independent locus at 3' of CDKAL1 and novel variants of several loci with type 2 diabetes in Chinese*, Diabetologia, 56(12):2619-2628, December, 2013. (e-published 9/8/2013; doi: 10.100/S0013-013-3047-1) PMID: 24013783 PMCID: 3825282

2. Luan, B., Goodarzi, M.O., Phillips, N.G., Guo, X., Chen, Y.D.I., Yao, J., Allison, M., Rotter, J.I., Shaw, R., Montminy, M.: *Macrophage class IIa HDACs link cAMP signaling to inflammation and insulin resistance*, Cell Metabolism, 19:1-8, June 3, 2014. (e-published 4/22/2014, doi:10.016/j.cmet.2014.03.024) PMID: 24768298 PMCID: 4207085

3. Wessel, J., Chu, A.Y., Willems, S… Garcia, M.E., Guo, X., Hara, K… Strawbridge, R.J., Taylor, K.D., Bork-Jensen, J… Chen, Y., Chen, Y.-D.I., Correa, A… Dupuis, J., Rotter, J.I., Meigs, J.B., Scott, R.A., Goodarzi, M.O.: *Low-frequency and rare exome chip variants associate with fasting glucose and type 2 diabetes susceptibility - The CHARGE T2D-Glycemia Exome Consortium*, Nature Communications, 6:5897, January 29, 2015 (doi:10.1038/ncomms6897) PMID: 25631608 PMCID: 4311266

4. Knowles, J.W., Xie, W., Zhang, Z… Morris, A., Chen, Y.-D.I., Mäkinen, V.-P., Ganna, A., Guo, X., Abassi, F… Schadt, E.E., Rotter, J.I., Sinaiko, A… Weedon, M.N., Walker, M., Quertermous, T.: *Identification and validation of NAT2 as an insulin sensitive gene*, Journal of Clinical Investigation, 125(4):1739-1751, April 1, 2015. (e-published 3/23/2015, doi:10.1172/JCI74692) PMID: 25798622 PMCID: 4409020

5. Palmer, N.D., Goodarzi, M.O., Langefeld, C.D., Wang, N., Guo, X., Taylor, K.D., Norris, J.M… Kandeel, F., Chen, Y.-D.I., Bowden, D.W… Raffel, L.J., Rotter, J.I., Watanabe, R.M., Wagenknecht, L.E.: *Genetic variants associated with quantitative glucose homeostasis traits translate to type 2 diabetes in Mexican Americans: The GUARDIAN (Genetics Underlying Diabetes in Hispanics) Consortium*, Diabetes, 64(5):1853-1866, May, 2015. (e-published 12/18/2014) PMID: 25524916

6. Liu, C.-T., Raghavan, S., Maruthur, N… Gauton, K.J., Guo, X., Hayes, M.G… Rybin, D.V., Taylor, K.D., Agyemang, C… Langefeld, C.D., Li, X., Liang, J… Arnett, D.K., Chen, Y.-D.I., Nalls, M.A… Florez, J.C., Rotter, J.I., Morris, A.P., Meigs, J.B.: *Trans-ethnic meta-analysis illuminates the genetic architecture of fasting glucose and insulin*, American Journal of Human Genetics, 99:56-75, July 7, 2016 (http://dx.doi.org/10.1016/j.ajhg.2016.05.006) PMID: 27321945

7. Qi, Q., Stilp, A.M., Sofer, T… MEDIA Consortium, Chen, Y.-D.I., Taylor, K.D., Aviles-Santa, M.L… Laurie, C.C., Rotter, J.I., Kaplan, R.C.: *Genetics of diabetes in U.S. Hispanic/Latino individuals: results from the Hispanic Community Health Study/Study of Latinos (HCHS/SOL)*, Diabetes, 66(5):1419-1425, May, 2017. (e-published 3/2/2017, doi:10.2337/db16-1150) PMID: 28254843 PMCID: 5399610

8. Wheeler, E., Leong, A., Liu, C.-T… Chasman, D.I., Chen, Y.-D.I., Chen, Y.-T… Soranzo, N., Guo, X., Roberts, D.J… Selvin, E., Rotter, J.I., Langenberg, C., Barroso, I., Meigs, J.B.: *Impact of common genetic determinants of Hemoglobin A1C on type 2 diabetes risk and diagnosis in ancestrally diverse populations: A transethnic genome-wide meta-analysis*, PLOS Medicine, 14(9):e1002383, September 12, 2017. (https://doi.org/10.1371/journal.pmed.1002383)

9. Zhao, W., Rasheed, A., Takkanen, E… Sheu, W.H.-H., Guo, X., Hsiung, C.A., Juang, J.-M., Taylor, K.D., Hung, Y.-J… Tai, E.S., Rotter, J.I., Chen, Y.-D.I., Frossard, P… Danesh, J., Voight, B., Saleheen, D.: *Seventeen novel loci associated with type 2 diabetes and shared etiological pathways with coronary heart disease*, Nature Genetics, 49(10):1450-1457, October, 2017. (e-published

9/4/2017, doi:10.1038/ng.3943) PMID: 28869590

10. Mahajan, A., Wessel, J., Willems, S.M… Rayner, N.W., Guo, X., Lu, Y… Burtt, N.P., Chen, Y.-D.I., Ching-Ti, L… Stančáková, A., Taylor, K.D., Teslovich, T.M…. Morris, A.P., Rotter, J.I., McCarthy, M.I.: *Refining the accuracy of validated target identification through coding variant fine-mapping in type 2 diabetes*, Nature Genetics, 50(4):559-571, April, 2018. (e-published 4/9/2018, doi:10.1038/s41588-018-0084) PMID: 29632382

11. Palmer, N.D., Okut, H., Hsu, F.-C., Ng, M.C.Y., Chen, Y.-D.I., Goodarzi, M.O., Taylor, K.D., Norris, J.M., Lorenzo, C., Rotter, J.I., Bergman, R.N., Langefeld, C.D., Wagenknecht, L.E., Bowden, D.W.: *Untargeted metabolomics identifies distinctive metabolite signatures for measures of glucose homeostasis: The Insulin Resistance Atherosclerosis Family Study (IRAS-FS)*, Journal of Clinical Endocrinology and Metabolism, 103:1877-1888, May, 2018. (doi:10.1210/jc.2017-02203) PMID: 29546329

12. Gusarova, V., O'Dushlaine, C., Teslovich, T.M… Thorsteinsdottir, U., Rotter, J.I., Chuang, L.-M… Rader, D.J., Chen, Y.-D.I., Hveem, K… Baras, A., Dewey, F.E., Gromada, J.: *Genetic inactivation of ANGPTL4 is associated with improved glucose homeostasis and reduced risk of type 2 diabetes*, Nature Communications, 9(1):2252, June 13, 2018. (doi:10.1038/s41467-018-04611-z) PMID: 29899519

13. Mahajan, A., Talium, D., Thurner, M… Prins, B.P., Guo, X., Bielak, L.F… Strauch, K., Taylor, K.D., Thorand, B… Meigs, J.B., Rotter, J.I., Marchini, J… Morris, A.P., Boehnke, M., McCarthy, M.I.: *Fine-mapping of an expanded set of type 2 diabetes loci to single-variant resolution using high-density imputation and islet-specific epigenome maps*, Nature Genetics, 50(11):1505-1513, November, 2018. (e-published 10/8/2018, doi:10.1038/s41588-018-0241-6) PMID: 30297969

14. Flannick, J., Mercader, J.M., Fuchsberger, C… Rich, S., Rotter, J.I., DiscovEHR Collaboration… Florez, J.C., McCarthy, M.I., Boehnke, M.: *Genetic discovery and translational decision support from exome sequencing of 20,791 type 2 diabetes cases and 24,440 controls from five ancestries*, Nature, 570(7759):71-76, June, 2019. (e-published 5/22/2019, doi: 10.1038/s41586-019-1231-2) PMID: 31118516

15. Leong, A., Chen, J., Wheeler, E… Tai, E.S., Rotter, J.I., Florez, J.C., Barroso, I., Meigs, J.B.: *Mendelian randomization analysis of hemoglobin A1c as a risk factor for coronary artery disease*, Diabetes Care, 42(7):1202-1208, July, 2019. (e-published 1/18/2019, pii: dc181712, doi:10.2337/dc18-1712) PMID: 30659074 PMCID: 6609962

16. Moon, J.-Y., Louie, T.L., Jain, D… Bottinger, E.P., Chen, Y.-D.I., Taylor, K.D., Daviglus, M… Schneiderman, N., Rotter, J.I., Kaplan, R.C., Qi, Q.: *A genome-wide association study identifies blood disorder related variants influencing hemoglobin A1C with implications for glycemic status in US Hispanics/Latinos*, Diabetes Care, 42(9):1784-1791, September, 2019. (e-published 6/18/2019, doi:10.2337/dc19-0168) PMID: 31213470 PMCID: 6702612

17. Goodarzi, M.O., Palmer, N.D., Cui, J., Guo, X., Chen, Y.-D.I., Taylor, K.D., Raffel, L.J., Wagenknecht, L.E., Buchanan, T.A., Hsueh, W.A., Rotter, J.I.: *Classification of type 2 diabetes genetic variants and a novel genetic risk score association with insulin clearance*, Journal of Clinical Endocrinology and Metabolism, 105(4):1251-1260, April 1, 2020. (e-published 11/12/2019, doi:10.1210/clinem/dgz198) PMID: 31714576 PMCID: 7059988

18. Spracklen, C.N., Horikoshi, M., Kim, Y.J… Chee, M.-L., Chen, Y.-D.I., Chen, Y.-T… Gross, M., Guo, X., Guo, Y… Cho, Y.S., Rotter, J.I., Wang, Y-X… Kim, B.-J., Mohlke, K.L., Sim, X.: *Identification of type 2 diabetes loci in 433,540 East Asian individuals*, Nature, 582(7811):240-245, June 6, 2020. (e-published 5/6/2020, doi.org/10.1038/s41586-020-2263-3) PMID: 32499647 PMCID: 7292783

B. Cardiovascular Disease (Includes CAD, Subclinical Atherosclerosis, Stroke, and Valvular Disease) (16 examples)

1. Ibrahim-Verbaas, C.A., Fornage, M., Bis, J.C… Rice, K., Rotter, J.I., Taylor, K., Folsom, A… Longstreth, W., van Duijn, C.M., Launer, L.J.: *Predicting stroke through genetic risk functions: the CHARGE Risk Score Project*, Stroke, 45(2):403-412, February, 2014. (e-published 1/16/2014, doi:10.1161/STROKEAHA.113.003044 PMID: 24436238 PMCID: 3955258

2. Smith, J.G., Luk, K., Schulz, C.-A… Budoff, M.J., Rotter, J.I., Cupples, L.A… O'Donnell, C.J., Post, W.S., Thanassoulis, G. for the Cohorts for Heart and Aging Research in Genetic Epidemiology (CHARGE) Extracoronary Calcium Working Group: *Genetic elevated low-density lipoprotein cholesterol is associated with aortic valve calcification and incident aortic stenosis*, Journal of the American Medical Association, 312(17):1764-1771, November 5, 2014. (e-published 10/26/2014, doi:10.1001/jama.2014.13959) PMID: 25344734 PMCID: 4280258

3. Stitziel, N.O., Won, H.-H., Morrison, A.C… Siscovick, D., Rotter, J.I., Hazen, S… Boerwinkle, E., Gabriel, S., Kathiresan, S.: *Inactivating mutations in NPC1L1 and protection from coronary heart disease*, New England Journal of Medicine, 371:2072-2082, November 27, 2014. (e-published 11/12/14, doi:10.1056/NEJMoa1405386) PMID: 25390462 NIHMSID: 650128

4. Assimes, T.L., Lee, I.T., Juang, J.-M., Wang, T.-D… Hsiung, C.A., Rotter, J.I., Sheu, W.H.-H., Chen, Y.-D.I., Taylor, K.D.: *Genetics of coronary artery disease in Taiwan: A cardiometabochip study by the TAICHI Consortium Study*, PLOS One, 11(3):e0138014, March 16, 2016. (doi:10.1371/journal.pone.0138014) PMID: 26982883 PMCID: 4794124

5. Howson, J.M.M., Zhao, W., Barnes, D… Lee, W.-J., Taylor, K.D., Chen, Y.-H., Lee, I.-T., Guo, X., Chung, R.-H., Hung, Y.-J., Rotter, J.I., Juang, J.-M.J... Chowdhury, R., Chen, Y.-D.I., Nordestgaard, B.G… Danesh, J., Butterworth, A.S., Saleheen, D.: *Fifteen new risk loci for coronary artery disease, highlighted arterial wall-specific mechanisms*, Nature Genetics, 49(7):1113-1119, July, 2017. (e-published 5/22/2017, doi: 10.1038/ng.3874) PMID: 28530674

6. Dewey, F.E., Gusarova, V., Dunbar, R.L… Liang, K.-W., Guo, X., Rotter, J.I., Chen, Y.-D.I., on behalf of the TAICHI consortium, Kraus, W.E… Mellis, S.J., Gromada, J., Baras, A.: *Genetic and pharmacologic inactivation of ANGPTL3 and atherosclerotic cardiovascular disease*, New England Journal of Medicine, 377(3):211, July 20, 2017. (e-published 5/24/2017, doi:10.1056/NEJMoa1612790) PMID: 28538136

7. Malik, R., Chauhan, G., Traylor, M… Rothwell, P.M., Rotter, J.I., Rundek, T… Kamatani, Y., Debette, S., Dichgans, M.: *Multi-ancestry genome-wide association study of 520,000 subjects identifies 32 loci associated with stroke and stroke subtypes*, Nature Genetics, 59(4):524-537, April, 2018. (e-published 3/12/208, doi:10.1038/s41588-018-0058-3) PMID: 29531354

8. Hajek, C., Guo, X., Jie, Y., Yang, H… Psaty, B.M., Taylor, K.D., Rotter, J.I.: *Coronary Heart Disease (CHD) Genetic Risk Score (GRS) predicts cardiovascular disease risk in men, not women, The Multi-Ethnic Study of Atherosclerosis,* Circulation Genomic and Precision Medicine, 11:e002324, October, 2018. (doi:10.1161/CIRCGEN.118.002324)

9. Khera, A.V., Chaffin, M., Zekavat, S.M… Spertus, .A., Taylor, K.D., Psaty, B.M… Lander, E., Chen, Y.D.I., Talkowski, M.E., Rotter, J.I., Krumholz, H.M., Kathiresan, S.: *Whole genome sequencing to characterize monogenic and polygenic contribution in patients hospitalized with early-onset myocardial infarction*, Circulation, 139(13):1593-1602, March 26, 2019. (epublished 11/11/2018, doi:10.1161/CIRCULATIONAHA.118.035658) PMID: 30586733

10. Agha, G., Mendelson, M.M., Ward-Caviness, C… Wiggins, K.L., Rotter, J.I., Malthaup, M., Hou, L., Levy, D., Baccarelli, A.A.: *Blood leukocyte DNA methylation predicts future risk of myocardial*

infarction and coronary heart disease: A study of 11,461 participants in nine population-based cohorts, Circulation, 140(8):645-657, August 20, 2019. (e-published 8/19/2019, doi:10.1161/CIRCULATIONAHA.118.039357) PMID: 31424985

11. Tzoulaki, I., Castagné, C.L., Karaman, I… Ferreira, D.L.S., Guo, X., Rotter, J.I., Taylor, K.D., Karvousi, M… Herrington, D., Lindon, J.C., Elliot, P.: *Serum metabolic signatures of coronary and carotid atherosclerosis and subsequent cardiovascular disease*, European Heart Journal, 40(34):2883-2896, September 7, 2019. (e-published 5/18/2019, doi:10.1093/eurheartj/ehz235) PMID: 31102408

12. Aung, N., Vargas, J.D., Manichaikul, A.W… Neubauer, S., Rotter, J.I., Lima, J., Taylor, K.D., Kawut, S., Munroe, P.B., Petersen, S.E.: *Genome-wide association study of left ventricular image-derived phenotypes identifies fourteen loci implicated in cardiogenesis and heart failure development*, Circulation, 140(16):1318-1330, October 15, 2019. (e-published 9/25/2019, doi: 10.1161/CIRCULATIONAHA.119.041161) PMID: 31554410.

13. Malhotra, R., Mauer, A.C., Guo, X., Yao, J… Criqui, M., Rotter, J., Johnson, A.D… Bloch, D.B., Post, W.S., O'Donnell, C.J., for the CHARGE Extracoronary Calcium Working Group: *HDAC9 is implicated in atherosclerotic aortic calcification and regulates vascular smooth muscle cell function*, Nature Genetics, 51(11):1580-1587, November, 2019. (e-published October 28, 2019, doi:10.1038/5415588-019-0514-8) PMID: 31659325 PMCID: 6858575

14. Mosley, J.D., Gupta, D.K., Tan, J….. Guo, X., Rotter, J.I., Roden, D.M., Gerszten, R.E., Wang, T.J.: *Predictive accuracy of a polygenic risk score compared with a clinical risk score for incident coronary heart disease*, The Journal of the American Medical Association, 323(7):627-635, February 18, 2020. (doi:10.1001/jama.2019.21782) PMID: 32068817

15. Goodarzi, M.O., Rotter, J.I.: *Genetic insights in the relationship between type 2 diabetes and coronary heart disease*, Circulation Research, 126(11):1526-1548, May 22, 2020. (e-published 5/21/2020, doi:10.1161/CIRCRESAHA.119.316065) PMID: 32437307 PMCID: 7250006

16. Rotter, J.I., Lin, H.J.: *An outbreak of polygenic scores for coronary artery disease*, Journal of the American College of Cardiology, 75(22):2781-4, June 9, 2020. (e-published 6/1/2020, doi.org/10.1016/j.jacc.2020.04.054)

C. Ocular Disorders (Includes Diabetic Retinopathy, Keratoconus, Mascular Degeneration, Glaucoma, Retinal Vasculature) (8 examples)

1. Sheu, W.H.-H., Kuo, J.Z., Lee, I.-T… Klein, B., Ipp, E., Lin, S.-Y., Guo, X., Hsieh, C.-H., Taylor, K.D., Fu, C.-P., Rotter, J.I., Chen, Y.-D.I.: *Genome-wide association study in a Chinese population with diabetic retinopathy*, Human Molecular Genetics, 22(15):3165-3173, August 1, 2013. (e-published 4/4/2013, doi: 10.1093/hmg/ddt161) PMID 23562823 PMCID: 3699066

2. Gao, X., Gauderman, J., Marjoram, P., Torres, M., Chen, Y.-D.I., Taylor, K.D., Rotter, J.I., Varma, R.: *Native American ancestry is associated with proliferative diabetic retinopathy in Latinos*, Investigative Opthalmology and Visual Science, 55(9):6041-6045, September, 2014. (e-published August 21, 2014, doi:10.1167/iovs.14-15044) PMID: 25146985 PMCID: 4176415

3. Kuo, J.Z., Guo, X., Klein, R… Goodarzi, M.O., Rotter, J.I., Chen, Y.-D.I., Ipp, E.: *Adiponectin, insulin sensitivity and diabetic retinopathy in Latinos with type 2 diabetes*, Journal of Clinical Endocrinology and Metabolism, 100(9):348-55, September, 2015. (e-published May 28, 2015, doi:10.1210/jc2015-1221) PMID: 26020763

4. Iglesias, A.I., Mishra, A., Vitart, V… van Leeuwen, E.M., Taylor, K.D., Blue Mountains Eye Study – GWAS group, Bonenmaijer, P., Rotter, J.I., Martin, N.G…. Burdon, K.P., van Duijn, C.M., MacGregor, S.: *Understanding the genetic architecture of central corneal thickness and its role in related blinding disorders in a genome-wide cross-ancestry meta-analysis*, Nature Communications, 9(1):1864, May 14, 2018. (doi:10.1038/s41467-018-03646-6) PMID: 29760442 PMCID: 5951816

5. Zangwill, L.M., Ayyagari, R., Liebmann, J.M… Christopher, M.A., Chen, Y.-D.I., Guo, X., Taylor, K.D., Rotter, J.I., Weinreb, R.N. for ADAGES III Genomics Study Group: *The African descent and glaucoma evaluation study (ADAGES) III: Contribution of genotype to glaucoma phenotype in African Americans. Study design and baseline data*, Opthalmology, 126(1):156-170, January, 2019. (e-published 1/17/2018, pii:S0161-6420(17)32714-9, doi: 10.1016/j.ophtha.2017.11.031) PMID: 29361356 PMCID: 6050158

6. Taylor, K.D., Guo, X., Zangwill, L.M… Ng, M.C.Y., Chen, Y.-D.I., Ayyagari, R., Rotter, J.I., Weinreb, R.N. for the ADAGES III Genomics Study Group: *Genetic architecture of POAG in individuals of African descent: The African Descent & Glaucoma Evaluation Study (ADAGES) III*, Opthalmology, 126(1):38-48, January, 2019. (e-published 10/20/2018, doi:10.1016/j.ophtha.2018.10.031) PMID: 30352225

7. Pollack, S., Igo, R.P., Jensen, R.A., Christiansen, M., Li, X., Cheng, C.-Y… Tan, G., Chen, Y.-D.I., Kuo, J.Z… Park, K.H., Guo, X., Ipp, E.K., Taylor, K.D., Adler, S… Klein, R., Rotter, J.I., Iyengar, S., Price, A., Sobrin, L.: *Multi-ethnic genome-wide association study of the diabetic retinopathy using liability threshold modeling of duration and diabetes and glycemic control*, Diabetes, 68(2):441-456, February, 2019. (e-published 11/28/2018, doi:10.2337/db18-0567) PMID: 30487263 PMCID: 6341299

8. Hauser, M.A., Allingham, R.R., Aung, T… Van Der Heide, C.J., Taylor, K.D., Rotter, J.I., Wang, S.-H.J... Challa, P., Chen, Y.-D.I., Chuka-Okosa, C.M… Guirou, N., Guo, X., Haines, J.L… Koch, A.E.A., Fingert, J.H., Khor, C.C.: *APBB2 is significant risk locus for primary open angle glaucoma in individuals of African ancestry*, Journal of the American Medical Association, 322(17):1682-1691, November 5, 2019. (doi:10.1001/JAMA.2019.16161)

D. Lipids (8 examples)

1. Global Lipids Genetics Consortium, Willer, C.J., Schmidt, E.M., Sengupta, S… Kleber, M.E., Li, X., Luan, J… Chambers, J.C., Chen, Y-D.I., Collins, F.S… Chasman, D.I., Rotter,J.I., Franks, P.W… Mohlke, K.L., Ingelsson, E., Abecasis, G.R.: *Discovery and refinement of loci associated with lipid levels*, Nature Genetics, 45(11):1274-1283, November, 2013. (e-published 10/1/2013; doi:10.1038/ng.2797). PMID: 24097068 PMCID: 3838666

2. Do, R., Willer, C.J., Schmidt, E.M… Kleber, M.E., Li, X., Luan, J… Chambers, J.C., Chen, Y.-D.I., Collins, F.S… Chasman, D.I., Rotter, J.I., Franks, P.W… Daly, M.J., Neale, B., Kathiresan, S.: *Common variants associated with plasma triglycerides and risk for coronary artery disease*, Nature Genetics, 45(11):1345-1352, November, 2013. (e-published 10/6/2013; doi: 10.1038ng.2795) PMID: 24097064 PMCID: 3904346

3. Peloso, G.M., Auer, P.L., Bis, J.C… Harris, T., Taylor, K.D., Andrews, J.S… Levy, D., Chen, Y.-D.I., Reiner, A.P… Psaty, B.M., Rotter, J.I., O'Donnell, C.J… Boerwinkle, E., Kathiresan, S., Cupples, L.A.: *Association of a low-frequency and rare protein-coding sequence variants with blood lipids in 56,000 individuals of European and African-American Ancestries*, American Journal of Human Genetics, 94(2):223-232, February 6, 2014. (dx.doi.org/10.1016/j.ajhg.2014.01.009) PMID: 24507774 PMCID: 3928662

4. Liu, D.J., Peloso, G.M., Yu, H… Chen, Y.E., Chen, Y.-D.I., Chowdhury, R… Roden, D.M., Rotter, J.I., Rudan, I… Tardif, J.-C., Taylor, K.D., Trompet, S… Musunuru, K., Willer, C.J., Kathiresan, S.: *Exome-wide association study of plasma lipids in >300,000 individuals*, Nature Genetics, 49(12):1758-1766, December, 2017. (e-published 10/30/2017, doi:10.1038/ng.3977) PMID: 29083408 PMCID: 5709146

5. Lu, X., Peloso, G.M., Liu, D.J… Long, J., Guo, X., Xu, M., Spracklen, C.N., Chen, Y., Liu, X… Sham, P.C., Gu, D., Willer, C.J.: *Exome chip meta-analysis identifies novel loci and East Asian-specific coding variants contributing to lipid levels and coronary artery disease*, Nature Genetics, 49(12):1722-1730, 2017. (e-published 10/30/2017, doi:10.1038/ng.3978) PMID: 29083407

6. Natarajan, P., Peloso, G.M., Zekavat, S.M… Cupples, A., Rotter, J.I., NHLBI TOPMed Lipids Working Group, Willer, C.J., Kathiresan, S.: *Deep-coverage whole genome sequences and blood lipids among 16,324 individuals*, Nature Communications, 9(1):3391, August 23, 2018. (doi:10.1038/s41467-018-05747-8) PMID: 30140000 PMCID: 6107638

7. Bentley, A.R., Sung, Y.J., Brown, M.R… Deng, X., Guo, X., Liu, J., Chen, G., Chen, Y.-D.I., Christensen, K… Tang, H., Taylor, K.D., Teo, Y.Y… Franceschini, N., Rotter, J.I., Zhu, X… Rao, D.C., Rotimi, C.N., Cupples, L.A.: *Multi-ancestry genome-wide association study incorporating gene x smoking interactions identifies novel lipid loci*, Nature Genetics, 51(4):636-648, April, 2019. (e-published 3/29/2019, doi:10.1038/s41588-019-0378-y) PMID: 30926973

8. Li, X., Li, Z., Zhou, H… Freedman, B.I., Guo, X., Kardia, S.L.R…. NHLBI Trans-Omics for Precision Medicine (TOPMed) Consortium Lipids Working Group, Rotter, J.I., Willer, C., Peloso, G.M., Natarajan, P., Lin, X.: *Dynamic incorporation of multiple-in-silico functional annotations empowers rare variant association analysis in large-scale whole genome sequencing studies*, Nature Genetics, 52(9):969-983, September, 2020. (e-published 8/24/2020, doi:10.1038/s4158-020-0676-4) PMID: 32839606

E. Blood Pressure and Hypertension (6 examples)

1. Liu, C., Kraja, A.T., Smith, J.A… Weiss, S., Guo, X., Palmas, W., Martin, L.W., Chen, Y.-D.I., Surendran, P… O'Donnell, C., Taylor, K.D., Liu, K… Chakravarti, A., Rotter, J.I., Psaty, B.M… Newton-Cheh, C., Levy, D., Chasman, D.I.: *Meta-analysis identifies common and rare variants influencing blood pressure and overlapping with metabolic trait loci*, Nature Genetics, 48(10):1162-70, October, 2016. (e-published 9/12/2016, doi:10.1038/ng.3660) PMID: 27618448

2. Ehret, G.B., Ferreira, T., Chasman, D.I… Kleber, M.E., Guo, X., Lyytikäinen, L.-P… Hovingh, G.K., Taylor, K.D., Sedoussis, G… Salomaa, V., Chen, Y.-D.I., Rotter, J.I., Froguel, P… Morris, A.P., Newton-Cheh, C., Munroe, P.B.: *The genetics of blood pressure regulation and its target organs from association studies in 342,415 individuals*, Nature Genetics, 48(10):1171-84, October, 2016. (e-published 9/12/2016, doi:10.1038/ng.3667) PMID: 27618452 PMCID: 5042863

3. Liang, J., Le, T.H., Edwards, D.R.V… Palmas, W., Guo, X., Papanicolaou, G.J… Rao, D.C., Rotter, J.I., Rotimi, C.. Chakravarti, A., Zhu, X., Franceschini, N.: *Single and multi-trait genome wide association analyses identify novel loci for blood pressure in African-ancestry populations*, PLOS Genetics, 13(5):e1006728, May 12, 2017. (doi:10.1371/journal.pgen.1006728) PMID: 2848854

4. Richard, M.A., Huan, T., Ligthart, S… Psaty, B.M., Chen, Y.-D.I., Kardia, S.L.R… Damcott, C.M., Rotter, J.I., Palmas, W… O'Connell, J.R., Guo, X., Pausova, Z., Dehghan, A., Levy, D., Fornage, M.: *DNA methylation analysis identifies novel loci for blood pressure regulation*, American Journal of Human Genetics, 101:1-15, December 7, 2017. (doi:10.1016/j.ajhg.2017.09.028) PMID: 29198723

5. Evangelou, E., Warren, H.R., Mosen-Ansorena, D… Gudnason, V., Guo, X., Gyllensten, U… Rose, L.M., Rotter, J.I., Rudan, I… Swertz, M., Taylor, K., Teumer, A… Wain, L.V., Elliott, P., Caufield, M.J.: *Genetic analysis over one million people identifies 535 novel loci for blood pressure*, Nature Genetics, 50(10):1412-1425, October, 2018. (e-published 9/17/2018, doi:10.1038/s41588-018-0205-x) PMID: 30224653

6. Surendran, P., Feofanova, E.V., Lahrouchi, N... Gudbjartsson, D.F., Guo, X., Harris, S.E… Chambers, J.C., Chen, Y.-D.I., Chowdhury, R… Rosendaal, F.R., Rotter, J.I., Rudan, I… Sylvain, S., Taylor, K.D., Timo, L.A… Newton-Cheh, C., Munroe, P.B., Howson, J.M.M.: *88 rare variants associated with blood pressure regulation in meta-analysis of ~1.3 million individuals*, Nature Genetics, in press, 2020.

F. EKG and Arrhythmias (8 examples)

1. den Hoed, M., Eijgelsheim, M., Esko, T… Quertermous, T., Rotter, J.I., van der Schouw, V.T… Snieder, H., Samani, N.J. Loos, R.J.F.: *Identification of heart rate-associated loci and their effects on cardiac conduction and rhythm disorders*, Nature Genetics, 45(6):621-631, June, 2013. (e-published 4/14/2013, doi:10.1038/ng.2610) PMCID: 3696959

2. Arking, D.E., Pulit, S., Crotti, L., van der Harst, P… Psaty, B., Rotter, J.I., Kolcic, I… Pfeufer, A., de Bakker, P.I.W., Newton-Cheh, C.: *Genetic association study of QT interval highlights calcium signaling pathways in myocardial repolarization*, Nature Genetics, 46(8):826-836, 2014. (e-published 6/22/14, doi:10.1038/ng.3014) PMID: 24952745 PMCID: 4124521

3. van der Harst, P., van Setten, J., Verweij, N… Ric, K.M., Rotter, J.I., Schlessinger, D… Isaacs, A., Samani, N.J., deBakker, P.I.W.: *Fifty-two genetic loci influencing human electrically active myocardial mass*, Journal of American College of Cardiology, 68(13):1435-48, September 27, 2016. (doi:10.1016/j.jacc.2016.07.729) PMID: 27659466

4. Lubitz, S.A., Yin, X., Lin, H.J… Furie, K.L., Guo, X., Larson, M.G… Stott, D.J., Taylor, K.D., Weng, L.-C… Rosand, J., Rotter, J.I., Heckbert, S.R., Ellinor, P.T., Alonso, A., Benjamin, E.J. on behalf of the AFGen Consortium: *Genetic risk prediction of atrial fibrillation*, Circulation, 135(14):13111320, April 4, 2017. (e-published 10/28/2016, pii: CIRCULATIONAHA.116.024143) PMID: 27793994 PMCID: 5380584

5. Christophersen, I., Lunetta, K.L., Yin, X… Chen, L.Y., Chen, Y.-D.I., Sandhu, R.K… Anderson, C.D., Taylor, K.D., Campbell, A… Sinner, M.F., Lin, H.J., Guo, X., METASTROKE Consortium of the ISGC… Hayward, C., Rotter, J.I., März, S… Rienstra, M., Lubitz, S.A., Ellinor, P.T., on behalf of the AFGen Consortium: *Common and rare variant analyses in >150,000 individuals reveal twelve novel genetic loci associated with atrial fibrillation*, Nature Genetics, 49(6):946-952, June, 2017. (e-published 4/17/2017, doi:10.1038/ng.3843) PMID: 28416818

6. Roselli, C., Chaffin, M.D., Weng, L.-C… Chen, L.Y., Chen, Y.-D.I., Choi, E.-K… Gudnason, V., Guo, X., Gupta, N… Lin, H., Lin, H.J., Lind, L… Rost, N., Rotter, J.I., Saba, S….. Lubitz, S.A., Lunetta, K.L., Ellinor, P.T.: *Genome-wide association analyses for atrial fibrillation reveals 70 novel loci*, Nature Genetics, 50(9):1225-1233, November 21, 2018. (e-published 6/11/2018, doi:10.1038/s41588-018-0133-9) PMID: 29892015

7. Ashar, F.N., Mitchell, R.N., Albert, C.M… McKnight, B., Rotter, J.I., Lemaitre, R.N… Siscovick, D.S., Arking, D.E., Sotoodehnia, N., for the SCD working group of the CHARGE Consortium: *A comprehensive evaluation of the genetic architecture of sudden cardiac arrest*, European Heart Journal, 39(44):3961-3969, November 21, 2018. (e-published, 8/28/2018, doi:10.1093/eurheartjehy474) PMID: 30169657 PMCID: 6247663

8. Khera, A.V., Mason-Suares, H., VanDenburgh, M.J… Gaziano, J.M., Taylor, K.D., Sotoodehnia, N… Rich, S.S., Rotter, J.I., Lander, E.S… Lebo, M., Albert, C.M., Kathiresan, S.: *Gene sequencing for adult-onset sudden cardiac death*, Journal of the American College of Cardiology, 74(21):2623-2634, November 26, 2019. (doi:10.1016/j.jacc.219.08.160) PMID: 31727422

G. Pharmacogenetics (5 examples)

1. Mangravite, L.M., Engelhardt, B.E., Medina, M.W… Rieder, M.J., Chen, Y.-D., Rotter, J.I., Ridker, P.M… Nickerson, D.A., Stephens, M., Krauss, R.M.: *A statin-dependent QTL for GATM expression is associated with statin-induced myopathy*, Nature, 502(7471):337-380, October 17, 2013. (e-published 8/28/2013, doi:10.1038/nature12508) PMID: 23995691 PMCID: 3933266

2. Postmus, I., Trompet, S., Deshmukh, H.A., Barnes, M., Li, X., Warren, H., Chasman, D.I… Feng, Q., Taylor, K.D., Li, G… Shaw-Hawkins, S., Chen, Y.-D.I., Nickerson, D.A… Wilke, R.A., Guo, X., Heckbert, S.R… Palmer, C., Rotter, J.I., Cupples, L.A… Krauss, R.M., Jukema, J.W., Caulfield, M.J.: *Pharmacogenetic meta-analysis of genome-wide scans of LDL cholesterol response to statins*, Nature Communications, 5:5068, October 28, 2014. (doi:10.1038/ncomms6068) PMID: 25350695 PMCID: 4220464

3. Scott, R.A., Freitag, D.F., Li, L., Chu, A.Y… Stančáková, A., Chen, Y., Varga, T… Riboli, E., Rotter, JI., Saleheen, D… Ehm, M.G., Wareham, N.J., Waterworth, D.M.: *Genomic approach to therapeutic target validation identifies a glucose-lowering GLPIR variant protective for coronary heart disease*, Science Translational Medicine, 8(341):341ra76, June 1, 2016. (doi:10.1126/scitranslmed.aad3744) PMID: 27252175

4. Smit, R.A.J., Postmus, I., Trompet, S., Barnes, M.R., Warren, H., Arsenault, B.J., Chasman, D.I., Cupples, L.A., Hitman, G., Krauss, R.M., Li, X., Psaty, B.M., Stein, C.M., Rotter, J.I., Jukema, J.W., also on behalf of the GIST consortium investigators: *Rooted in risk; genetic predisposition for LDL-c levels associates with diminished LDL-c response to statin treatment*, Pharmacogenomics Journal, 17(15):1621-1628, 2016. (e-published 9/20/2016, doi:10.2217/pgs-2016-0091) PMID: 27648687

5. Wei, W.-Q., Li, X., Feng, Q… Wilke, R.A., Rotter, J.I., Stein, C.M… Roden, D., Krauss, R.M., Denny, J.C.: *LPA gene variants are associated with residual cardiovascular risk in patients receiving statins*, Circulation, 138(17):1839-1849, October 23, 2018. (e-published 4/27/2018, doi:1161/CIRCULATIONAHA.117.031356) PMID: 29703846 PMCID: 6202211

H. Anthropometry (Includes Adiposity and Height) (7 examples)

1. Monda, K.L., Chen, G.K., Taylor, K.C… Chen, F., Chen, Y.-D.I., Chiang, C.W.K… Gottesman, O., Guo, X., Haritunians, T… Loos, R.J.F., North, K.E., Haiman, C.A.: *A meta-analysis identifies new loci associated with body mass index in individuals of African ancestry*, Nature Genetics, 45(6):690-6, June, 2013. (e-published 4/14/2013, doi:10.1038/ng.2607) PMID: 23583978 PMCID: 3694490

2. Wood, A.R., Esko, T., Yang, J… Rivadeneria, F., Rotter, J.I., Saaristo, T.E… Visscher, P.M., Frayling, T., Hirschhorn, J.: *Defining the role of common variation in the genomic and biological architecture of adult human height*, Nature Genetics 45(11):1173-1186, November, 2014. (e-published 10/5/2014) PMID: 25282103 PMCID: 4250049

3. Locke, A., Kahali, B., Berndt, S… Caspersen, I., Chen, Y.-D., Clarke, R… Hirschhorn, J., Loos, R., Speliotes, E.K.: *Genetic studies of body mass index yield new insights for obesity biology*, Nature, 518(7538):197-206, February 12, 2015. (doi:10.1038/nature14177) PMID: 25673413 PMCID: 4382211

4. Chu, A.Y., Deng, X., Fisher, V.A… Gudnason, V., Guo, X., Harris, T.B… Mosely Jr., T.H., Rotter, J.I., Shuldiner, A.R… Cupples, L.A., Steinhauser, M.L., Fox, C.S.: *Multiethnic genome-wide meta-analysis of ectopic fat deposits identifies loci associated with adipocyte development and differentiation*, Nature Genetics, 49(1):125-130, January, 2017. (e-published 12/5/2016, doi:10.1038/ng.3738) PMID: 27918534

5. Marouli, E., Graff, M., Medina-Gomez, C… Mahajan, A., Guo, X., Abecasis, G… Chasman, D.I., Chen, Y.-D.I., Chowdhury, R… Tardif, J.-C., Taylor, K.D., Teumer, A… MAGIC Investigators, Rotter, J.I., Boehnke, M… Hirschhorn, J.N., Deloukas, P., Lettre, G.: *Rare and low-frequency coding variants alter human adult height*, Nature, 542(7640):186-190, February 9, 2017. (e-published 2/1/2017, doi:10.1038/nature21039) PMID: 28146470 PMCID: 5302847

6. Turcot, V., Lu, Y., Highland, H.M… Graff, M., Guo, X., Hendricks, A.E… Chasman, D.I., Chen, Y.-D.I., Chowdhury, R… Tardif, J.-C., Taylor, K.D., Teumer, A… Understanding Society Scientific Group, Rotter, J.I., Pospisilik, J.A… Lindgren, C.M., Hirschhorn, J.N., Loos, R.J.F.: *Protein altering variants associated with body mass index implicate pathways that control energy intake and expenditure underpinning obesity*, Nature Genetics, 50:26-41, January, 2018.

7. Justice, A.E., Karaderi, T., Highland, H.M… Fine, R.S., Guo, X., Schurmann, C… Chasman, D.I., Chen, Y.-D.I., Chowdhury, R… Tardif, J.-C., Taylor, K.D., Teumer, A… Mohlke, K.L., Rotter, J.I., Kutalik, Z… Loos, R.J.F., North, K.E., Lindgren, C.M.: *Protein-coding variants highlight the importance of lipolysis in adipocytes for body fat distribution*, Nature Genetics, 51(3):452-469, March, 2019. (e-published 2/18/2019, doi:10.1038/s41588-018-0334-2) PMID: 30778226

I. Genomics of Selected Other Common Disorders (Includes Neurologic, Pulmonary, Reproductive, Hematologic, Renal) (15 examples)

1. Lambert, J.-C., Ibrahim-Verbaas, C.A., Harold, D… Bettens, K., Rotter, J.I., Brice, A… Williams, J., Schellenberg, G.D., Amouyel, P.: *Meta-analysis of 74,046 individuals identifies 11 new susceptibility loci for Alzheimer's disease*, Nature Genetics, 45(12):1452-1458, December, 2013. (e-published 10/27/2013; doi:10.1038/ng.2802) PMID: 24162737 PMCID: 3896259

2. Loth, D.W., Artigas, M.S., Garib, S.A… Morrison, A.C., Rotter, J.I., Gao, W… , Brusselle, G.G., Tobin, M.D., London, S.J.: *Genome-wide association analysis identifies six new loci associated with forced vital capacity*, Nature Genetics, 46(7):669-677, July, 2014. (e-published 6/15/2014, doi:10.1038/ng.3011) PMID: 24929828 PMCID: 4140093

3. van der Lee, S.J., Holstege, H., Wong, T.H… Emilsson, V., Rotter, J.I., Aspelund, T… van der Flier, W.M., Ikram, A., van Duijn, C.M.: PLD3-variants in population studies, Nature, 520:E2-E3, April 2, 2015. (doi:10.1038/nature14038)

4. Day, F.R., Ruth, K.S., Thompson, D.J… Chapman, R., Chen, Y.-D.I., Chenevix-Trench, G… Guénel, P., Guo, X., Haiman, C.A… Langenberg, C., Li, X., Lindström, S… Swerdlow, A., Taylor, K.D., Thorsteindottir, U… Ridker, P.M., Rotter, J.I., Toniolo, D.D… Murabito, J.M., Perry, J.R.B.B., Murray, A.: *Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair*, Nature Genetics, 47(11):1294-303, November, 2015. (e-published 9/28/2015, doi:10.1038/ng.3412) PMID: 26414677 PMCID: 4661791

5. CHARGE Consortium Hematology Working Group, Pankratz, N., Schick, U.M… Rivadeneira, F., Rotter, J.I., Starr, J.M., Taylor, K.D., Thuesen, B.H… O'Donnell, C.J., Reiner, A.P., Ganesh, S.K., on behalf of the CHARGE Hematology Working Group: *Meta-analysis of rare and common exome chip variants identifies S1PR4 and other novel genes influencing blood cell traits*, Nature Genetics, 48(8):867-76, August, 2016. (e-published 7/11/2016, doi:10.1038/ng.3607) PMID: 27399967

6. Okbay, A., Baselmans, B.M., De Neve, J.E… Tanaka, T., Taylor, K., Wedenoja, J… Benjamin, D.J., Bartels, M., Cesarini, D.: *Genetic variants associated with subjective well-being, depressive symptoms, and neuroticism identified through genome-wide analysis*, Nature Genetics 40(5):404-15, July, 2016. (e-published 4/18/2016, doi:10.1038/ng.3552) PMID: 27089181 (Corrigendum, 48(12):1591, November 29, 2016, doi:10.1039/ng1216-1587b) PMID: 27898078

7. Barban, N., Jansen, R., de Vlaming, R… Gunderson, E.P., Guo, X., Hayward, C… den Hoed, M., Snieder, H., Mills, M.C.: *Genome-wide analysis identifies 12 loci influencing human reproductive behavior*, Nature Genetics, 48(12):1462-1472, December, 2016. (e-published 10/31/2016; doi:10.1038/ng.3698) PMID: 27798627

8. Sims, R., van der Lee, S.J., Naj, A.C… Leber, M., Chen, Y., Albert, M.S… Munger, R., Rotter, J.I., Lipton, R.B… Seshadri, S., Williams, J., Schellenberg, G.D.: *Rare coding variants in PLCG2, AB13 and TREM2 implicate microglial-mediated innate immunity in Alzheimer's disease*, Nature Genetics, 49(9):1373-1384, September, 2017. (e-published 7/17/2017, doi:10.1038/ng.3916) PMID: 28714976 PMCID: 5669039

9. Anttila, V., Bulik-Sullivan, B., Finucane, H.K… Riedel-Heller, S., Rotter, J.I., Ransmayr, G… Rosan, J., Corvin, A., Neale, B.M.: *Analysis of shared heritability in common disorders of the brain*, Science, 350(6395), June 22, 2018. (pii: eaap8757, doi:10.1126/science.aap8757) PMID: 29930110.

10. Kunkle, B.W., Grenier-Boley, B., Vronskaya, M… Larson, E.B., Rotter, J.I., Bullido, M.J…

Schellenberg, G.D., Lambert, J.-C., Pericak-Vance, M.A.: *Genetic meta-analysis of diagnosed Alzheimer's disease identifies new risk loci and implicates Aβ, tau, immunity and lipid processing*, Nature Genetics, 51(3):414-430, March, 2019. (e-published 2/28/2019, doi:10.1038/s41588-019-0358-2) PMID: 30820047

11. Wuttke, M., Li, Y., Li, M… Tan, N.Y.Q., Taylor, K., Teren, A… Zonderman, A.B., Rotter, J.I., Bochud, M… Teumer, A., Köttgen, A., Pattaro, C.: *A catalogue of molecular targets for kidney function from genetic analyses of a million individuals*, Nature Genetics, (6):957-972, June, 2019. (e-published 5/31/2019, doi:10.1038s41588-019-0407-x) PMID: 31152163

12. Satizabal, C.L., Adams, H.H.h… Hegenscheid, K., Rotter, J.I., Schork, A.J… Thompson, P.M., Seshadri, S., Ikram, M.A.: *Genetic architecture of subcortical brain structures in 38,844 individuals worldwide*, Nature Genetics, 51(11):1624-1636, November, 2019. (e-published 10/21/2019, doi: 10.1038/s41588-018-0511-γ) PMID: 31636452

13. Bick, A.G., Weinstock, J.S., Nandakumar, S.K... de Andrade, M., Guo, X., Konkle, B.A…. McGarvey, S.T., Chen, Y.-D.I., Shoemaker, B... Desai, P., Taylor, K.D., Johnson, A… Rich, S.S., Rotter, J.I., Wilson, J.G... Sankaran, V.G., Natarajan, P., Kathiresan, S., on behalf of the NHLBI Trans-Omics for Precision Medicine (TOPMed) Consortium: *Inherited causes of clonal hematopoiesis of indeterminate potential in TOPMed whole genomes*, Nature, 586(7831):763-768, October, 2020. (e-published 10/14/2020, doi:10.1038/s41586-020-2819-2)

14. Chen, M.-H., Raffield, L.M., Mousas, A… Rosen, J.D., Rotter, J.I., Schubert, P… Reiner, A.P., Auer, P.L., Lettre, G.: *Trans-ethnic and ancestry-specific blood-cell genetics in 746,667 individuals from 5 global populations*, Cell182(5):1198-1213.e14, September 3, 2020. (doi:10.1016/j.cell.2020.06.045) PMID: 32888493

15. Vuckovic, D., Bao, E.L., Akbari, P… Rosen, J.D., Rotter, J.I., Schubert, P… Lettre, G., Sankaran, V.G., Soranzo, N.: *The polygenic and monogenic basis of blood traits and diseases*, Cell, 182(5):1214-1231.e11, September 3, 2020. (doi:10.1016/j.cell.2020.08.008) PMID: 32888494

J. Population Genetics (Includes Population Genetics, Genetic Epidemiology, Statistical Genetics, Risk Assessment, Mitochondrial Disorders, and Miscellanous Disorders) (9 examples)

1. Grove, M.L., Yu, B., Cochran, B.J… Bis, J.C., Taylor, K.D., Hansen, M… O'Donnell, C.J., Rotter, J.I., Boerwinkle, E.: *Best practices and joint calling of the Human Exome Bead Chip: the CHARGE Consortium*, PLOS One, 8(7):e68095, July 12, 2013 (doi:10.1371/journal.pone.0068095) PMID: 23874508 PMCID: 3709915

2. Amendola, L.M., Dorschner, M.O., Robertson, P.D… Lee, K., Taylor, K.D., Guo, X., Crooks, K… Yang, Y., Rotter, J.I., Rich, S.S... Rehm, H., Nickerson, D.A., Jarvik, G.P.: *Actional exomic incidental findings in 6503 participants: challenges of variant classification*, Genome Research, 25(3):305-315, March, 2015. (e-published 1/30/2015) PMID: 25637381 PMCID: 4352885

3. Joshi, P.K., Esko, T., Mattsson, H… Trompet, S., Guo, X., Chasman, D.I… Chen, C., Chen, Y.-D.I., Collins, F.S… Tang, S.-T., Taylor, K.D., Tayo, B.O… Ridker, P.M., Rotter, J.I., Sattar, N… Polašek, O., Wilson, J.F., Wilson, for ROHgen: *Evidence for directional dominance on complex traits relating to size and cognition in a wide range of human populations*, Nature, 523(7561):459-462, July 23, 2015. (e-published 7/1/2015, doi: 10.1038/nature14618) PMID: 26131930 PMCID: 4516141

4. Conomos, M.P., Laurie, C.A., Stilp, A… Avery, C., Taylor, K.D., Rotter, J.I., Talavera, G… Rice, K., Thornton, T., Laurie, C.C.: *Genetic diversity and association studies in U.S. Hispanic/Latino populations: applications in the Hispanic Community Health Study/Study of Latinos, American Journal of Human Genetics*, 98(1):165-84, January 7, 2016. (doi:10.1016/j.ajhg.2015.12.001) PMID: 26748518

5. Brody, J.A., Morrison, A., Bis, J.C… Rich, S., Rotter, J.I., Wilson, J., Boerwinkle, E., Psaty, B.M., Cupples, L.A.: *Analysis commons: Team science in a big-data environment for genetic epidemiology*, Nature Genetics, 49(11):1560-1563, October 27, 2017. (doi:10.1038/ng.3968) PMID: 29074945

6. Jones, L., Wells, K., Lin, H.J… Wright, A., Young, K., Rotter, J.I.: *Community partnership in precision medicine: Themes from a community engagement conference*, Ethnicity and Disease, 28(Suppl 2): 503-510, 2018. (doi:10.18865/ed.28.S2.503)

7. Guo, X., Rotter, J.I.: *Genome-wide association studies,* Journal of the American Medical Association, 322(17):1705-1706, November 5, 2019.

8. Gutierrez-Arcelus, M., Baglaenko, Y., Arora, J… Rich, S.S., Taylor, K.D., Rotter, J.I., Esko, T., Brenner, M.B., Raychaudhuri, S.: *Allele-specific expression changes dynamically during T-cell activation in HLA and other autoimmune loci*, Nature Genetics, 52(3):247-253, March, 2020. (e-published 2/17/20, doi: 10.1038/s41588-020-0579-4) PMID: 32066938 PMCID: 7135372

9. Collins, R.L., Brand, H., Karczewski, K.J… Spertus, J.A., Taylor, K.D., Lin, H.J., Rich, S.S., Post, W., Chen, Y.-D.I., Rotter, J.I., Nusbaum, C… Banks, E., MacArthur, G., Talkowski, M.E.: *A structural variation reference for medical and population genetics*, Nature, 581(7809):444-451, May 28, 2020. (epublished 5/27/2020, doi:10.1038/s41586-020-2287-8) PMID: 32461652 PMCID: 7334194