Biographical and Bibliographic Information

JOSHUA W. KNOWLES

I. Identifying data:

Professional Address: Falk CVRC, CV 273
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II. Academic history:

Colleges and universities attended, degrees received, dates.

8/91-5/95	BA	History, Biology (w/*honors*) University of North Carolina-Chapel Hill, NC
8/95-5/03	MD	Medicine University of North Carolina-Chapel Hill, NC
6/97-8/01	PhD	Genetics and Molecular Biology University of North Carolina-Chapel Hill, NC

Scholarships and honors (pre-MD degree)

1991-1993	Freshman and Sophomore Honors Program, UNC-Chapel Hill, NC
1991-1995	Dean's List several semesters, UNC-Chapel Hill, NC

Post-doctoral and residency training

7/1/03-6/30/05	Internship and Residency, Internal Medicine Stanford University School of Medicine, Stanford, CA
7/1/05-6/30/10	Cardiology Fellow, Division of Cardiovascular Medicine Stanford University School of Medicine, Stanford, CA

Medical Board eligibility or boards passed, with date(s) (include licensure)

8/2006	A.B.I.M. Certification, Internal Medicine
10/2010	A.B.I.M. Certification, Cardiovascular Disease (recertified 2019)
11/2012	Diplomate, American Board of Clinical Lipidology

Licensure

7/2004-	California Medical License	# A87974
10/2007-	Drug Enforcement Agency License	# FK0548175

III. Employment history. List all academic and non-academic positions.

Academic positions:

3/2023-Present	Associate Professor of (Cardiovascular) Medicine (University Medical Line)
7/2020-8/2024	Faculty Affiliate, Stanford Prevention Research Center
9/2018-8/2024	Reappointed Sept 1, 2018 Assistant Professor of (Cardiovascular) Medicine (Medical Center Line)
9/2014-9/2018	Initial appointment, Assistant Professor of (Cardiovascular) Medicine, Medical Center Line
7/2011-8/2014	Instructor, Division of Cardiovascular Medicine Stanford University School of Medicine, Stanford, CA
8/2010-6/2011	Clinical Instructor, Division of Cardiovascular Medicine, Stanford University School of Medicine, Stanford, CA

Non-Academic positions: None

IV. Honors and Awards

1995	Graduated with Honors, double major in History and Biology, UNC
1996	John B. Graham Medical Student Research Society, UNC School of Medicine
2003-2004	Stanford Clinical Investigator Pathway for Internal Medicine Residents
2005-2006	Stanford University School of Medicine Dean's Fellowship
2006-2008	AHA Postdoctoral Fellowship Award
2006	The Future Leaders in CV Medical Research Program, Fellowship Award
2007, 2008	Edwin Alderman Award for Clinical Research, Stanford Cardiovascular Medicine
2010-2015	AHA National Fellow-to-Faculty Award
2012	Elected Fellow, American Heart Association
2012	Elected Fellow, American College of Cardiology
2015	ACCF/Herman K. Gold Young Investigator's Award Finalist in Molecular/Cellular Cardiology
2016-2019	Clinical Scientist Award, Doris Duke Charitable Trust
2019	Elected Fellow, National Lipid Association

V. Scholarly Publications

A. Peer reviewed journal articles

1. Li Z, **Knowles JW**, Goyeau D, Prabhakar S, Short DB, Perkins AG, Goy MF. Low salt intake down-regulates the guanylin signaling pathway in rat distal colon. *Gastroenterology* 1996; 111(6):1714-1721.
2. **Knowles JW**, Reddick RL, Jennette JC, Shesely EG, Smithies O, Maeda N. Enhanced atherosclerosis and kidney dysfunction in eNOS(-/-)Apoe(-/-) mice are ameliorated by enalapril treatment. *J Clin Invest* 2000; 105(4):451-458.

3. **Knowles JW**, Esposito G, Mao L, Hagaman JR, Fox JE, Smithies O, Rockman HA, Maeda N. Pressure-independent enhancement of cardiac hypertrophy in natriuretic peptide receptor A-deficient mice. *J Clin Invest* 2001; 107(8):975-984.
4. Goy MF, Oliver PM, Purdy KE, **Knowles JW**, Fox JE, Mohler PJ, Qian X, Smithies O, Maeda N. Evidence for a novel natriuretic peptide receptor that prefers brain natriuretic peptide over atrial natriuretic peptide. *Biochem J* 2001; 358(Pt 2):379-387.
5. Ellmers LJ, **Knowles JW**, Kim HS, Smithies O, Maeda N, Cameron VA. Ventricular expression of natriuretic peptides in Npr1(-/-) mice with cardiac hypertrophy and fibrosis. *Am J Physiol Heart Circ Physiol* 2002; 283(2):H707-714.
6. Hodgin JB, **Knowles JW***, Kim HS, Smithies O, Maeda N. Interactions between endothelial nitric oxide synthase and sex hormones in vascular protection in mice. *J Clin Invest* 2002; 109(4):541-548.
7. **Knowles JW**, Erickson LM, Guy VK, Sigel CS, Wilder JC, Maeda N. Common variations in noncoding regions of the human natriuretic peptide receptor A gene have quantitative effects. *Hum Genet* 2003; 112(1):62-70.
8. Alexander MR, **Knowles JW***, Nishikimi T, Maeda N. Increased atherosclerosis and smooth muscle cell hypertrophy in natriuretic peptide receptor A-/-apolipoprotein E-/- mice. *Arterioscler Thromb Vasc Biol* 2003;23(6):1077-1082.
9. Caron KM, James LR, Kim HS, **Knowles J**, Uhlir R, Mao L, Hagaman JR, Cascio W, Rockman H, Smithies O. Cardiac hypertrophy and sudden death in mice with a genetically clamped renin transgene. *Proc Natl Acad Sci U S A* 2004; 101(9):3106-3111.
10. **Knowles JW**, Wang H, Itakura H, Southwick A, Myers RM, Iribarren C, Fortmann SP, Go AS, Quertermous T, Hlatky MA. Association of polymorphisms in platelet and hemostatis system genes with acute myocardial infarction. *Am Heart J* 2007 154(6):1052-1058.
11. Assimes TL, **Knowles JW**, Priest JR, Basu A, Borchert A, Volcik KA, Grove ML, Tabor HK, Southwick A, Tabibiazar R, Sidney S, Boerwinkle E, Go AS, Iribarren C, Hlatky MA, Fortmann SP, Myers RM, Kuhn H, Risch N, Quertermous T. A near null variant of 12/15-LOX encoded by a novel SNP in ALOX15 and the risk of coronary artery disease. *Atherosclerosis* 2008; 198(1):136-144.
12. **Knowles JW***, Assimes TL, Boerwinkle E, Fortmann SP, Go A, Grove ML, Hlatky M, Iribarren C, Li J, Myers R, Risch N, Sidney S, Southwick A, Volcik KA, Quertermous T. Failure to replicate an association of SNPs in the oxidized LDL receptor gene (OLR1) with CAD. *BMC Med Genet* 2008 Apr; 9:23.
13. Assimes TL, **Knowles JW***, Priest JR, Basu A, Volcik KA, Southwick A, Tabor HK, Hartiala J, Allayee H, Grove ML, Tabibiazar R, Sidney S, Fortmann SP, Go A, Hlatky M, Iribarren C, Boerwinkle E, Myers R, Risch N, Quertermous T. Common polymorphisms of ALOX5 and ALOX5AP and risk of coronary artery disease. *Hum Genet* 2008; 123(4): 399-408.
14. Assimes TL, **Knowles JW**, Basu A, Iribarren C, Southwick A, Tang H, Absher D, Li J, Fair JM, Rubin GD, Sidney S, Fortmann SP, Go AS, Hlatky MA, Myers RM, Risch N, Quertermous T. Susceptibility locus for clinical and subclinical coronary artery disease at chromosome 9p21 in the multi-ethnic ADVANCE Study. *Hum Mol Genet* 2008; 17(15): 2320-2328.
15. Zakharia F, Basu A, Absher D, Assimes TL, Go AS, Hlatky MA, Iribarren C, **Knowles JW**, Li J, Narasimhan B, Sidney S, Southwick A, Myers RM, Quertermous T, Risch N,

Tang H. Characterizing the admixed African ancestry of African Americans. *Genome Biol* 2009; 10(12):R141.

16. Ingelsson E, [10 authors], **Knowles JW**, [54 authors], Florez JC; MAGIC Investigators [42 inst]. Detailed physiologic characterization reveals diverse mechanisms for novel genetic Loci regulating glucose and insulin metabolism in humans. *Diabetes* 2010; 59(5):1266-1275. [genotyping and data analysis]
17. Dackor J, Tobacco & Genetics Consortium; Franceschini N, ARIC; Bernardinelli L, ATVB Italian Study Group; **Knowles JW**, ADVANCE; [114 auth, 103 inst]. Genome-wide meta-analyses identify multiple loci associated with smoking behavior. *Nat Genet* 2010 May; 42(5):441-447. [exchange of genotyping data, assistance writing manuscript].
18. Ashley EA, Butte AJ, Wheeler MT, Chen R, Klein TE, Dewey FE, Dudley JT, Ormond KE, Pavlovic A, Morgan AA, Pushkarev D, Neff NF, Hudgins L, Gong L, Hodges LM, Berlin DS, Thorn CF, Sangkuhl K, Hebert JM, Woon M, Sagreiya H, Whaley R, **Knowles JW**, Chou MF, Thakuria JV, Rosenbaum AM, Zaranek AW, Church GM, Greely HT, Quake SR, Altman RB. Clinical assessment incorporating a personal genome. *Lancet* 2010 May; 375(9725):1525-1535. [assistance with certain design questions, writing manuscript and critical review]
19. Lango Allen H, [51 authors], **Knowles JW**, [239 authors], Hirschhorn JN; GIANT Consortium. Hundreds of variants clustered in genomic loci and biological pathways affect human height. *Nature* 2010 Oct; 467(7317):832-838. [exchange of genotyping data, assistance writing manuscript]
20. Speliotes EK, [136 authors], **Knowles JW**, [18 authors], Ludwig B; MAGIC, Manunta P, [153 authors], Watkins H; Procardis Consortium, Wilson JF, [33 authors], Loos RJ. Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. *Nat Genet* 2010 Nov; 42(11):937-948. [collaboration through exchange of genotyping data, assistance writing manuscript]
21. Heid IM, [39 authors], Wood AR; MAGIC, Estrada K, [24 authors], **Knowles JW**, [232 authors], Lindgren CM. Meta-analysis identifies 13 novel loci associated with waist-hip ratio and reveals sexual dimorphism in the genetic basis of fat distribution. *Nature Genet* 2010 Nov; 42(11):949-960. [exchange of genotyping data]
22. Assimes TL, [14 authors], **Knowles JW**, [83 authors], Ball SG; Myocardial Infarction Genetics Consortium; Wellcome Trust Case Control Consortium; Cardiogenics, Ouwehand WH, [29 authors], Quertermous T. Lack of association between the Trp719Arg polymorphism in kinesin-like protein 6 and coronary artery disease in 19 case-control studies. *J Am Coll Cardiol* 2010 Nov;56(19):1552-1563. Erratum in: *J Am Coll Cardiol* 2011 Jan;57(4):520. [exchange of genotyping data, assistance writing manuscript]
23. Reilly MP*, Li M*, [22 authors], **Knowles J**, [25 authors]; †Wellcome Trust Case Control Consortium, Knouff CW, Waterworth DM, Walker MC, Martinelli N, Olivieri O, Girelli D, Quyyumi AA, Muhlestein JB, Anderson JL; †Myocardial Infarction Genetics Consortium, Erdmann J, Schunkert H, Hall AS, Quertermous T, Blankenberg S, Hazen SL, Roberts R, McPherson R, Kathiresan S, Samani NJ, Mooser V, Wilensky R, Hakonarson H, Epstein SE, Rader DJ. Identification of ADAMTS7 as a novel locus for coronary atherosclerosis and association of ABO with myocardial infarction in the presence of coronary athero-sclerosis: two genome-wide association studies. *Lancet* 2011 Jan;377(9763):383-392. [exchange of genotyping data]

24. Schunkert H, [70 authors], **Knowles JW,** [93 authors], Erdmann J; CARDIoGRAM Consortium, Samani NJ [> 100 authors]. Large-scale association analysis identifies 13 new susceptibility loci for coronary artery disease. *Nat Genet* 2011 Mar;43(4):333-338. [Contributed reagents/exchange of genotyping data, review of manuscript]
25. Speliotes EK, [24 authors]; NASH CRN [87 authors]; GIANT Consortium, [149 authors], **Knowles JW**, [183 authors]; MAGIC Investigators [273 authors]; GOLD Consortium [24 authors]. Genome-wide association analysis identifies variants associated with nonalcoholic faty liver disease that have distinct effects on metabolic traits. PLoS Genet 2011 Mar;7(3): e1001324. [14 pps.] [Contributed reagents/ materials/analysis tools]
26. Kraja AT, Vaidya D, Pankow JS, Goodarzi MO, Assimes TL, Kullo IJ, Sovio U, Mathias R, Sun YV, Franceschini N, Absher D, Li G, Zhang Q, Feitosa MF, Glazer NL, Haritunians T, Hartikainen AL, **Knowles JW**, North KE, Iribarren C, Kral B, Yanek L, O'Reilly PF, McCarthy MI, Jaquish C, Couper DJ, Chakravarti A, Psaty BM, Becker LC, Province MA, Boerwinkle E, Quertermous T, Palotie L, Jarvelin MR, Becker DM, Kardia SL, Rotter JI, Chen YD, Borecki IB. A bivariate genome-wide approach to metabolic syndrome: STAMPEED Consortium. *Diabetes* 2011 Apr; 60(4):1329-1339. [Helped with design, contributed reagents, exchange of genotyping data, assistance writing manuscript]
27. IBC 50K CAD Consortium; CARDIoGRAM Consortium. Schunkert H, [68 authors], **Knowles JW**, [91 authors], Samani NJ. Large-scale gene-centric analysis identifies novel variants for coronary artery disease. *PLoS Genet* 2011 Sep; 7(9):e1002260.
28. Dewey FE, Chen R, Cordero SP, Ormond KE, Caleshu C, Karczewski KJ, Whirl-Carrillo M, Wheeler MT, Dudley JT, Byrnes JK, Cornejo OE, **Knowles JW**, Woon M, Sangkuhl K, Gong L, Thorn CF, Hebert JM, Capriotti E, David SP, Pavlovic A, West A, Thakuria JV, Ball MP, Zaranek AW, Rehm HL, Church GM, West JS, Bustamante CD, Snyder M, Altman RB, Klein TE, Butte AJ, Ashley EA. Phased whole-genome genetic risk in a family quartet using a major allele reference sequence. *PLoS Genet* 2011 Sep; 7(9):e1002280. [Critical review of manuscript, assistance with certain design elements]
29. Heid IM, (67 authors) **Knowles JW**, Kraft P, (> 120 authors), North KE, O'Connell JR, Peltonen L, Schlessinger D, Strachan DP, Hirschhorn JN, Assimes TL, Wichmann HE, Thorsteinsdottir U, van Duijn CM, Stefansson K, Cupples LA, Loos RJ, Barroso I, McCarthy MI, Fox CS, Mohlke KL, Lindgren CM. Meta-analysis identifies 13 new loci associated with waist-hip ratio and reveals sexual dimorphism in the genetic basis of fat distribution. *Nat Genet* 2011 Oct 27;43(11):1164. doi: 10.1038/ng1111-1164a. [Contributed reagents/exchange of genotyping data, assistance writing manuscript]
30. Witteles RM***, Knowles JW***, Perez M, Morris WM, Spettell CM, Brennan TA, Heidenreich PA. Overuse of left ventriculography. *Am Heart J* 2012 Apr;163(4): 617-623.
31. **Knowles JW**, Assimes TL, Kiernan M, Pavlovic A, Goldstein BA, Yank V, McConnell MV, Absher D, Bustamante C, Ashley EA, Ioannidis JP. Randomized trial of personal genomics for preventive cardiology: design and challenges. *Circ Cardiovasc Genet* 2012 June; 5(3):368-376.
32. **Knowles JW**, Assimes TL, Tsao PS, Natali A, Mari A, Quertermous T, Reaven GM, Abbasi F. Measurement of insulin-mediated glucose uptake: Direct comparison of the modified insulin suppression test and the euglycemic, hyperinsulinemic clamp.

Metabolism 2012 Nov; doi:pii:S0026-0495(12)00384-8. 10.1016/j.metabol.2012.10.0002.

33. Xie W, Wood AR, Lyssenko V, Weedon MN, **Knowles JW**, Alkayyali S, Assimes TL, Quertermous T, Abbasi F, Paananen J, Häring H, Hansen T, Pedersen O, Smith U, Laakso M, Dekker JM, Nolan JJ, Groop L, Ferrannini E, Adam KP, Gall WE, Frayling TM, Walker M. Genetic variants associated with glycine metabolism and their role in insulin sensitivity and type 2 diabetes. *Diabetes* 2013 Feb 1. PMID: 23378610. [Contributed reagents/genotyping data, assistance writing manuscript].
34. Xie W, Fall T, Hao K, Ärnlöv J, Abbasi F, Schadt EE, Boran G, Hansen T, Greenawalt D, Nolan JJ, Pedersen O, Häring H, Ferrannini E, Syvänen AC, Quertermous T, Smith U, Assimes TL, Laakso M, Walker M, **Knowles JW**, Weedon MN, Frayling TM, Ingelsson E, on behalf of the GENESIS consortium. Mendelian randomization studies do not support a causal effect of plasma lipids on insulin sensitivity. *Diabetes* (*in press*, 2012). [Helped with design, writing manuscript, contributed data]
35. Dimas AS*, Lagou V*, Barker A*, **Knowles JW***, Mägi R, Hivert MF, Benazzo A, Rybin D, Jackson AU, Stringham HM, Song C, Fischer-Rosinsky A, Boesgaard TW, Grarup N, Abbasi F, Assimes TL, Hao K, Yang X, Lecoeur C, Barroso I, Bonnycastle LL, Böttcher Y, Bumpstead S, Chines PS, Erdos MR, Graessler J, Kovacs P, Morken MA, Narisu N, Payne F, Stancakova A, Swift AJ, Tönjes A, Bornstein SR, Cauchi S, Froguel P, Meyre D, Schwarz P, Boehnke M, Bergman RN, Collins FS, Mohlke KL, TuolimehtoJ, Quertemous T, Lind L, Hansen T, Pedersen O, Walker M, Pfeiffer AFH, Spranger J, Stumvoll M, Meigs JB, Wareham NJ, Kuusisto J, Laakso M, Langenberg C, Dupuis J, Watanabe RM*, Florez JC*, Ingelsson E*, McCarthy MI*, Prokopenko I*. Impact of loci contributing to type 2 diabetes susceptibility on variation in physiologic glycemic traits in healthy individuals. *Diabetes, 2013* PMID: 24296717. Epub ahead of print
36. CARDIoGRAMplusC4D Consortium, Deloukas P, Kanoni S, Willenborg C, Farrall M, Assimes TL, [45 authors]; DIAGRAM Consortium; CARDIOGENICS Consortium, [16 authors], **Knowles JW**, [15 authors]; MuTHER Consortium, [15 authors]; Wellcome Trust Case Control Consortium, [42 authors], Quertermous T, [43 authors], Samani NJ. Large-scale association analysis identifies new risk loci for coronary artery disease. *Nat Genet* 2013 Jan;45(1):25-33. [Contributed reagents/genotyping data, assistance writing manuscript]
37. Liang P, Lan F, Lee AS, Gong T, Sanchez-Freire V, Wang Y, Diecke S, Sallam K, **Knowles JW**, Nguyen PK, Wang PJ, Bers DM, Robbins RC, Wu JC. Drug screening using a library of human induced pluripotent stem cell-derived cardiomyocytes reveals disease specific patterns of cardiotoxicity. *Circulation* 2013 Mar; PMID: 23519760. [Recruited subjects and obtained biological specimens]
38. Coram MA, Duan Q, Hoffmann TJ, Thornton T, **Knowles JW**, Johnson NA, Ochs-Balcom HM, Donlon TA, Martin LW, Eaton CB, Robinson JG, Risch NJ, Zhu X, Kooperberg C, Li Y, Reiner AP*, Tang H*. Genome-wide characterization of shared and distinct genetic components that influence blood lipid levels in human populations. *ASHG* (*in press*). [Helped with specific aspects of design and with preparation of manuscript].

39. den Hoed M, Eijgelsheim M, Esko T (> 100 authors), **Knowles JW**, (> 100 authors) Snieder H, Samani NJ, Loos RJ. Identification of heart rate-associated loci and their effects on cardiac conduction and rhythm disorders. Nat Genet. 2013 Apr 14. doi: 10.1038/ng.2610. [Epub ahead of print] [Contributed data].
40. Hammond E, Watts GF, Rubinstein Y, Farid W, Livingston M, **Knowles JW**, Lochmüller H, Bellgard M, Dawkins HJ. Role of international registries in enhancing the care of familial hypercholesterolaemia. Int J Evid Based Healthc. 2013 Jun; 11(2):134-9.
41. Kuo JZ, Sheu WH, Assimes TL, Hung YJ, Absher D, Chiu YF, Mak J, Wang JS, Kwon S, Hsu CC, Goodarzi MO, Lee IT, **Knowles JW**, Miller BE, Lee WJ, Juang JM, Wang TD, Guo X, Taylor KD, Chuang LM, Hsiung CA, Quertermous T, Rotter JI, Chen YD. Trans-ethnic fine mapping identifies a novel independent locus at the 3' end of CDKAL1 and novel variants of several susceptibility loci for type 2 diabetes in a Han Chinese population. Diabetologia. 2013 Sep 8 [Epub ahead of print]. [Contributed reagents/genotyping data, assistance writing manuscript]
42. Yaghootkar H, Lamina C, Scott RA, Dastani Z, Hivert MF, Warren LL, Stancáková A, Buxbaum SG, Lyytikäinen LP, Henneman P, Wu Y, Cheung CY, Pankow JS, Jackson AU, Gustafsson S, Zhao JH, Ballantyne CM, Xie W, Bergman RN, Boehnke M, El Bouazzaoui F, Collins FS, Dunn SH, Dupuis J, Forouhi NG, Gillson C, Hattersley AT, Hong J, Kähönen M, Kuusisto J, Kedenko L, Kronenberg F, Doria A, Assimes TL, Ferrannini E, Hansen T, Hao K, Häring H, **Knowles** JW, Lindgren CM, Nolan JJ, Paananen J, Pedersen O, Quertermous T, Smith U; GENESIS Consortium; RISC Consortium, Lehtimäki T, Liu CT, Loos RJ, McCarthy MI, Morris AD, Vasan RS, Spector TD, Teslovich TM, Tuomilehto J, van Dijk KW, Viikari JS, Zhu N, Langenberg C, Ingelsson E, Semple RK, Sinaiko AR, Palmer CN, Walker M, Lam KS, Paulweber B, Mohlke KL, van Duijn C, Raitakari OT, Bidulescu A, Wareham NJ, Laakso M, Waterworth DM, Lawlor DA, Meigs JB, Richards JB, Frayling TM. Mendelian randomization studies do not support a causal role for reduced circulating adiponectin levels in insulin resistance and type 2 diabetes. Diabetes 2013 Oct; 62(10):3589-98. [Contributed reagents/genotyping data, assistance writing manuscript]
43. Heidenreich PA, Lin S, **Knowles JW**, Perez M, Maddox TM, Ho MP, Rumsfeld JS, Sahay A, Massie BM, Tsai TT, Witteles RM. Variation in use of left ventriculography in the veterans affairs health care system. Circ Cardiovasc Qual Outcomes. 2013 Nov 1; 6(6):687-93. doi: 10.1161/CIRCOUTCOMES.113.000199. Epub 2013 Nov 5. [helped initiate project, helped with writing manuscript]
44. Finocchiaro G, **Knowles JW**, Pavlovic A, Perez M, Magavern E, Sinagra G, Haddad F, Ashley E. Prevalence and Clinical Correlates of Right Ventricular Dysfunction in Patients With Hypertrophic Cardiomyopathy. Am J Cardiol. 2013 Oct 4. doi:pii: S0002-9149(13)01981-4. 10.1016/j.amjcard.2013.09.045. [Epub ahead of print] PMID: 24230980 [provided direction on the project, helped to write and edit manuscript]
45. Finocchiaro G, Murphy D, Pavlovic A, Haddad F, Shiran H, Sinagra G, Ashley EA, **Knowles JW**. Unexplained Double-Chambered Left Ventricle Associated with Contracting Right Ventricular Aneurysm and Right Atrial Enlargement. Echocardiography. 2013 Dec 3. doi: 10.1111/echo.12467. [Epub ahead of print. PMID: 24299065]
46. Emily C. O'Brien; Matthew T. Roe, Elizabeth S. Fraulo; Eric D. Peterson; Christie M.

Ballantyne; Jacques Genest; Samuel S. Gidding; Emma Hammond; Linda C. Hemphill; Lisa C. Hudgins; Iris Kindt; Patrick M. Moriarty; Joyce Ross; James A. Underberg; Karol Watson; Dave Pickhardt; Daniel J. Rader; Katherine Wilemon; **Joshua W. Knowles**. Rationale and design of the familial hypercholesterolemia foundation CAscade SCreening for Awareness and DEtection of Familial Hypercholesterolemia registry. American Heart Journal, 2013 December 23; 167(3):342-349.

47. Greendale, K, Pariani, M, **Knowles, JW**, (08/2014 [Last Update 07/25/11]). Familial Hypercholesterolemia (FH). In: Rare Disease Database. Copyright 2014, National Organization for Rare Disorders (NORD). Available at http://www.rarediseases.org.
48. Goldstein BA, **Knowles JW**, Salfati E, Ioannidis JP, Assimes TL. Simple, standardized incorporation of genetic risk into non-genetic risk prediction tools for complex traits: coronary heart disease as an example. Front Genet. 2014 Aug 1; 5:254. doi: 10.3389/fgene.2014.00254. eCollection 2014.PMID: 25136350.
49. **Knowles JW**, O'Brien EC, Greendale K, Wilemon K, Genest J, Sperling LS, Neal WA, Rader DJ, Khoury MJ. Reducing the burden of disease and death from familial hypercholesterolemia: a call to action. Am Heart J. 2014 Dec; 168(6):807-11. doi: 10.1016/j.ahj.2014.09.001. Epub 2014 Sep 16. PMID: 25458642.
50. Yaghootkar H, Scott RA, White CC, Zhang W, Speliotes E, Munroe PB, Ehret GB, Bis JC, Fox CS, Walker M, Borecki IB, **Knowles JW**, Yerges-Armstrong L, Ohlsson C, Perry JR, Chambers JC, Kooner JS, Franceschini N, Langenberg C, Hivert MF, Dastani Z, Richards JB, Semple RK, Frayling TM. Genetic evidence for a normal-weight "metabolically obese" phenotype linking insulin resistance, hypertension, coronary artery disease, and type 2 diabetes. Diabetes. 2014 Dec; 63(12):4369-77. doi: 10.2337/db14-0318. Epub 2014 Jul 21. PMID: 25048195.
51. Finocchiaro G, Haddad F, Pavlovic A, Magavern E, Sinagra G, **Knowles JW**, Myers J, Ashley EA. How does morphology impact on diastolic function in hypertrophic cardiomyopathy? A single centre experience. BMJ Open. 2014 Jun 12; 4(6):e004814. doi: 10.1136/bmjopen-2014-004814 PMID: 24928584.
52. Finocchiaro G, Kobayashi Y, Magavern E, Zhou JQ, Ashley E, Sinagra G, Schnittger I, **Knowles JW**, Fearon WF, Haddad F, Tremmel JA. Prevalence and prognostic role of right ventricular involvement in stress-induced cardiomyopathy.J Card Fail. 2015 Feb 19. pii: S1071-9164(15)00043-3. doi:10.1016/j.cardfail.2015.02.001. [Epub ahead of print]PMID: 25704104.
53. Finocchiaro G, Haddad F, **Knowles JW**, Caleshu C, Pavlovic A, Homburger J, Shmargad Y, Sinagra G, Magavern E, Wong M, Perez M, Schnittger I, Myers J, Froelicher V, Ashley EA. Cardiopulmonary responses and prognosis in hypertrophic cardiomyopathy: a potential role for comprehensive noninvasive hemodynamic assessment. JACC Heart Fail. 2015 May; 3(5):408-18. doi: 10.1016/j.jchf.2014.11.011. Epub 2015 Apr 8. PMID: 25863972.
54. Bent RE, Wheeler MT, Hadley D, **Knowles JW**, Pavolvic A, Finocchiaro G, Haddad F, Salisbury H, Race S, Shmargad Y, Matheson GO, Kumar V, Saini D, Froelicher V, Euan Ashley E, Perez MV. Systematic Comparison of Digital Electrocardiograms From Healthy Athletes and Patients With Hypertrophic Cardiomyopathy. J Am Coll Cardiol. 2015;65(22):2462-2463. doi:10.1016/j.jacc.2015.03.559.
55. Fall T, Xie W, Poon W, Yaghootkar H, Magi R; **Knowles JW**, Lyssenko V, Weedon M, Frayling TM, Ingelsson E, on behalf of the GENESIS Consortium. Using Genetic

Variants to Assess the Relationship Between Circulating Lipids and Type 2 Diabetes. Diabetes 2015 Jul;64(7):2676-84. doi: 10.2337/db14-1710.

56. **Knowles JW**, Stone NJ, Ballantyne CM. Familial Hypercholesterolemia and the 2013 American College of Cardiology/American Heart Association Guidelines: Myths, Oversimplification, and Misinterpretation Versus Facts. Diabetes 2015 Aug;116(3):481-4 doi: 10.1016/amjcard2015.04.062. PMID:26043952.
57. Goldstein BA, **Knowles JW**, Salfati E, Ioannidis JP, Assimes TL. Corrigendum: Simple standardized incorporation of genetic risk into non-genetic risk prediction tools for complex traits coronary heart disease as an example. Front Genet. 2015 Jul:6:231 doi: 10.3389/fgene.2015.00231. PMID: 26217377.
58. Priest JR, Yang W, Reaven G, **Knowles JW**, Shaw GM. Maternal Midpregnancy GlucoseLevels and Risk of Congenital Heart Disease in Offspring. JAMA Pediatr. 2015 Dec 1;169(12):1112-6. doi: 10.1001/jamapediatrics.2015.2831.PMID: 26457543.
59. Gidding SS, Ann Champagne M, de Ferranti SD, Defesche J, Ito MK, **Knowles JW**, McCrindle B, Raal F, Rader D, Santos RD, Lopes-Virella M, Watts GF, Wierzbicki AS The Agenda for Familial Hypercholesterolemia: A Scientific Statement From the American Heart Association. American Heart Association. Atherosclerosis, Hypertension, and Obesity in the Young Committee of the Council on Cardiovascular Disease in the Young, Council on Cardiovascular and Stroke Nursing, Council on Functional Genomics and Translational Biology, and Council on Lifestyle and Cardiometabolic Health. Circulation. 2015 Dec;132(22):2167-92. doi: 10.1161. Epub 2015 Oct 28. PMID: 26510694.
60. **Knowles JW**, Xie W, Zhang Z, Chennamsetty I, Assimes TL, Paananen J, Hansson O, Pankow J, Goodarzi MO, Carcamo-Orive I, Morris AP, Chen YD, Mäkinen VP, Ganna A, Mahajan A, Guo X, Abbasi F, Greenawalt DM, Lum P, Molony C, Lind L, Lindgren C, Raffel LJ, Tsao PS, Schadt EE, Rotter JI, Sinaiko A, Reaven G, Yang X, Hsiung CA, Groop L, Cordell HJ, Laakso M, Hao K, Ingelsson E, Frayling TM, Weedon MN, Walker M, Quertermous T. Identification and validation of N-acetyltransferase 2 as an Insulin Sensitivity Gene. J Clin Invest. 2016 Jan 4;126(1):403. doi: 10.1172/JCI85921. Epub 2016 Jan 4. PMID: 26727231
61. Ahmad ZS, Andersen RL, Andersen LH, O'Brien EC, Kindt I, Shrader P, Vasandani C, Newman CB, deGoma EM, Baum SJ, Hemphill LC, Hudgins LC, Ahmed CD, Kullo IJ, Gidding SS, Duffy D, Neal W, Wilemon K, Roe MT, Rader DJ, Ballantyne CM, Linton MF, Duell PB, Shapiro MD, Moriarty PM, **Knowles JW**. US physician practices for diagnosing familial hypercholesterolemia: data from the CASCADE-FH registry. *J Clin Lipidol*. 2016 Sep-Oct;10(5):1223-9. doi: 10.1016/j.jacl.2016.07.011.PMID: 27678440.
62. Abbasi F, Kohli P, Reaven GM, **Knowles JW**. Hypertriglyceridemia: A simple approach to identify insulin resistance and enhanced cardio-metabolic risk in patients with prediabetes. *Diabetes Res Clin Pract*. 2016 Oct;120:156-61. doi: 10.1016/j.diabres.2016.07.024. PMID 27565692.
63. Caleshu C, Kasparian NA, Edwards KS, Yeates L, Semsarian C, Perez M, Ashley E, Turner CJ,**Knowles JW**, Ingles J. Interdisciplinary psychosocial care for families with inherited cardiovascular diseases. *Trends Cardiovasc Med*. 2016 Oct;26(7):647-53. doi: 10.1016/j.tcm.2016.04.010. PMID: 27256036. [critical revision]
64. Burridge PW, Li YF, Matsa E, Wu H, Ong SG, Sharma A, Holmström A, Chang AC, Coronado MJ, Ebert AD, **Knowles JW**, Telli ML, Witteles RM, Blau HM, Bernstein D,

Altman RB, Wu JC. Human induced pluripotent stem cell-derived cardiomyocytes recapitulate the predilection of breast cancer patients to doxorubicin-induced cardiotoxicity. *Nat Med*. 2016 May;22(5):547-56. doi: 10.1038/nm.4087. PMID: 27089514. [material support, critical revision]

65. deGoma EM, Ahmad ZS, O'Brien EC, Kindt I, Shrader P, Newman CB, Pokharel Y, Baum SJ, Hemphill LC, Hudgins LC, Ahmed CD, Gidding SS, Duffy D, Neal W, Wilemon K, Roe MT, Rader DJ, Ballantyne CM, Linton MF, Duell PB, Shapiro MD, Moriarty PM, **Knowles JW**. Treatment Gaps in Adults With Heterozygous Familial Hypercholesterolemia in the United States: Data From the CASCADE-FH Registry. *Circ Cardiovasc Genet*. 2016 Jun;9(3):240-9. doi: 10.1161/CIRCGENETICS.116.001381. PMID: 27013694.
66. Finocchiaro G, Haddad F, Kobayashi Y, Lee D, Pavlovic A, Schnittger I, Sinagra G, Magavern E, Myers J, Froelicher V, **Knowles JW**, Ashley E. Impact of Septal Reduction on Left Atrial Size and Diastole in Hypertrophic Cardiomyopathy. *Echocardiography*. 2016 May;33(5):686-94. doi: 10.1111/echo.13158. PMID: 26926154. [supervison, critical revision, acquisition of data]
67. Kohli P, **Knowles JW***, Sarraju A, Waters DD, Reaven G. Metabolic Markers to Predict Incident Diabetes Mellitus in Statin-Treated Patients (from the Treating to New Targets and the Stroke Prevention by Aggressive Reduction in Cholesterol Levels Trials). *Am J Cardiol*. 2016 Nov 1;118(9):1275-1281. doi: 10.1016/j.amjcard.2016.07.054. PMID 27614854. *co-first authors
68. Chennamsetty I, Coronado M, Contrepois K, Keller MP, Carcamo-Orive I, Sandin J, Fajardo G, Whittle AJ, Fathzadeh M, Snyder M, Reaven G, Attie AD, Bernstein D, Quertermous T, **Knowles JW**. Nat1 Deficiency Is Associated with Mitochondrial Dysfunction and Exercise Intolerance in Mice. *Cell Rep.* 2016 Oct 4;17(2):527-540. doi: 10.1016/j.celrep.2016.09.005. PMID: 27705799.
69. Rodriguez, F, Maron, DJ, **Knowles JW,** Virani, SS, & Heidenreich, PA. Association Between Intensity of Statin Therapy and Mortality in Patients With Atherosclerotic Cardiovascular Disease. *JAMA Cardiology.* 2016 Nov 9. doi 10.1001/jamacardio. 2016.4052. [concept and design, critical revision]
70. Rodriguez, F, Lin, S., Maron, DJ, **Knowles, JW**, Virani, SS, & Heidenreich, PA. Use of high-intensity statins for patients with atherosclerotic cardiovascular disease in the Veterans Affairs Health System: Practice impact of the new cholesterol guidelines. *American Heart Journal.* 2016 Dec;182:97-102. doi: 10.1016/j.ahj.2016.09.007. [concept and design, critical revision]
71. Carcamo-Orive I, Hoffman GE, Cundiff P, Beckmann ND, D'Souza SL, **Knowles JW**, Patel A, Papatsenko D, Abbasi F, Reaven GM, Whalen S, Lee P, Shahbazi M, Henrion MY, Zhu K, Wang S, Roussos P, Schadt EE, Pandey G, Chang R, Quertermous T, Lemischka I. Analysis of Transcriptional Variability in a Large Human iPSC Library Reveals Genetic and Non-genetic Determinants of Heterogeneity. *Cell Stem Cell.* 2016 Dec 2. doi: 10.1016/j.stem.2016.11.005. PMID: 28017796.[concept and design, obtaining funding, drafting manuscript, supervision, critical revision]
72. **Knowles JW**, Howard WB, Karayan L, Baum SJ, Wilemon KA, Ballantyne CM, Myers KD. Access to Non-Statin Lipid Lowering Therapies in Patients at High-Risk of Atherosclerotic Cardiovascular Disease. *Circulation.* 2017 Apr 26. pii: CIRCULATIONAHA.117.027705. doi: 10.1161/CIRCULATIONAHA.117.027705.

73. Reaven GM, **Knowles JW,** Leonard D, Barlow CE, Willis BL, Haskell WL, Maron DJ. Relationship between simple markers of insulin resistance and coronary artery calcification. *J Clin Lipidol*. 2017 Jun 6: pii: S1933-2874(17)30339-2. doi: 10.1016/j.jacl.2017.05.013. [design and concept, critical revisions]
74. **Knowles JW**, Zarafshar S, Pavlovic A, Goldstein BA, Tsai S, Li J, McConnell MV, Absher D, Ashley EA, Kiernan M, Ioannidis JPA, Assimes TL. Impact of a Genetic Risk Score for Coronary Artery Disease on Reducing Cardiovascular Risk: A Pilot Randomized Controlled Study. *Front Cardiovasc Med.* **2017** Aug 14;4:53. doi: 10.3389/fcvm.**2017**.00053. eCollection **2017**.PMID 28856136
75. Amrock SM, Duell PB, Knickelbine T, Martin SS, O'Brien EC, Watson KE, Mitri J, Kindt I, Shrader P, Baum SJ, Hemphill LC, Ahmed CD, Andersen RL, Kullo IJ, McCann D, Larry JA, Murray MF, Fishberg R, Guyton JR, Wilemon K, Roe MT, Rader DJ, Ballantyne CM, Underberg JA, Thompson P, Duffy D, Linton MF, Shapiro MD, Moriarty PM, **Knowles JW**, Ahmad ZS. Health disparities among adult patients with a phenotypic diagnosis of familial hypercholesterolemia in the CASCADE-FH patient registry. *Atherosclerosis*. 2017 Dec;267:19-26. [concept and design, obtaining funding, supervision, critical revision]
76. Helle EIT, Biegley P, **Knowles JW,** Leader JB, Pendergrass S, Yang W, Reaven GR, Shaw GM, Ritchie M, Priest JR. *J Pediatr*. First Trimester Plasma Glucose Values in Women without Diabetes are Associated with Risk for Congenital Heart Disease in Offspring.2017 Dec 13. Pii: S0022-3476(17)31457-9. Doi: 10.1016/j.jpeds.2017. 10.046.[concept and design, obtaining funding, critical revision]
77. Chen AS, Bent RE, Wheeler M, **Knowles JW**, Haddad F, Froelicher V, Ashley E, Perez MV.Large Q and S waves in lead III on the electrocardiogram distinguish patients with hypertrophic cardiomyopathy from athletes. *Heart*. **2018** Apr 21. pii: heartjnl-2017-312647. doi: 10.1136/heartjnl-2017-312647. PMID 29680808 [material support, critical revision]
78. Poon AK, Meyer ML, Reaven G, **Knowles JW**, Selvin E, Pankow JS, Couper D, Loehr L, Heiss G. Short-Term Repeatability of Insulin Resistance Indexes in Older Adults: The Atherosclerosis Risk in Communities Study. *J Clin Endocrinol Metab*. **2018** Jun 1;103(6):2175-2181. doi: 10.1210/jc.2017-02437. PMID: 29618016 [design and concept, critical revisions]
79. Chang YC, Hee SW, Lee WJ, Li HY, Chang TJ, Lin MW, Hung YJ, Lee IT, Hung KY, Assimes T, **Knowles JW**, Nong JY, Lee PC, Chiu YF, Chuang LM. Genome-wide scan for circulating vascular adhesion protein-1 levels: MACROD2 as a potential transcriptional regulator of adipogenesis. *J Diabetes Investig*. **2018** Jan 24. doi: 10.1111/jdi.12805. PMID: 29364582 [critical revisions]
80. Chen AS, Bent RE, Wheeler M, **Knowles JW**, Haddad F, Froelicher V, Ashley E, Perez MV Large Q and S waves in lead III on the electrocardiogram distinguish patients with hypertrophic cardiomyopathy from athletes. Heart. 2018 Apr 21. pii: heartjnl-2017-312647. doi: 10.1136/heartjnl-2017-312647. PMID: 29680808
81. Rao AS, Lindholm D, Rivas MA, **Knowles JW**, Montgomery SB, Ingelsson E. Large-Scale Phenome-Wide Association Study of *PCSK9* Variants Demonstrates Protection Against Ischemic Stroke. Circ Genom Precis Med. 2018 Jul;11(7):e002162. doi: 10.1161/CIRCGEN.118.002162. PMID: 29997226 [design and concept, critical revisions]

82. Rodriguez F, **Knowles JW**, Maron DJ, Virani SS, Heidenreich PA. Frequency of Statin Use in Patients With Low-Density Lipoprotein Cholesterol ≥190 mg/dl from the Veterans Affairs Health System. Am J Cardiol. 2018 Jun 7. pii: S0002-9149(18)31180-9. doi: 10.1016/j.amjcard.2018.05.008. PMID: 30055758 [design and concept, critical revisions]
83. Sturm AC, **Knowles JW***, Gidding SS, Ahmad ZS, Ahmed CD, Ballantyne CM, Baum SJ, Bourbon M, Carrié A, Cuchel M, de Ferranti SD, Defesche JC, Freiberger T, Hershberger RE, Hovingh GK, Karayan L, Kastelein JJP, Kindt I, Lane SR, Leigh SE, Linton MF, Mata P, Neal WA, Nordestgaard BG, Santos RD, Harada-Shiba M, Sijbrands EJ, Stitziel NO, Yamashita S, Wilemon KA, Ledbetter DH, Rader DJ; Convened by the Familial Hypercholesterolemia Foundation. Clinical Genetic Testing for Familial Hypercholesterolemia: JACC Scientific Expert Panel. *J Am Coll Cardiol*. 2018 Aug 7;72(6):662-680. doi: 10.1016/j.jacc.2018.05.044. PMID: 30071997 *co-first authors
84. Kim D, Li AA, Cholankeril G, Kim SH, Ingelsson E, **Knowles JW**, Harrington RA, Ahmed A. Trends in overall, cardiovascular and cancer-related mortality among individuals with diabetes reported on death certificates in the United States between 2007 and 2017. *Diabetologia*. 2019 Apr 22. doi: 10.1007/s00125-019-4870-9. [Epub ahead of print] PMID: 31011776 [critical revision]
85. Iacocca MA, Chora JR, Carrié A, Freiberger T, Leigh SE, Defesche JC, Kurtz CL, DiStefano MT, Santos RD, Humphries SE, Mata P, Jannes CE, Hooper AJ, Wilemon KA, Benlian P, O'Connor R, Garcia J, Wand H, Tichy L, Sijbrands EJ, Hegele RA, Bourbon M, **Knowles JW**. *Hum Mutat*. 2018 Nov;39(11):1631-1640.PMID:30311388
86. Raygor V, Abbasi F, Lazzeroni LC, Kim S, Ingelsson E, Reaven GM, **Knowles JW.** Impact of race/ethnicity on insulin resistance and hypertriglyceridaemia. *Diab Vasc Dis Res*. 2019 Mar;16(2):153-159. PMID: 31014093
87. Rodriguez F, Maron DJ, **Knowles JW**, Virani SS, Lin S, Heidenreich PA. Association of Statin Adherence With Mortality in Patients With Atherosclerotic Cardiovascular Disease. *JAMA Cardiol*. 2019 Feb 13. PMID: 30758506 [concept and design, critical revision]
88. Harati H, Zanetti D, Rao A, Gustafsson S, Perez M, Ingelsson E, **Knowles JW**. No evidence of a causal association of type 2 diabetes and glucose metabolism with atrial fibrillation. *Diabetologia*. 2019 May;62(5):800-804. Epub 2019 Feb 27. PMID: 30810766
89. Tikkanen E, Gustafsson S, **Knowles JW**, Perez M, Burgess S, Ingelsson E. Body composition and atrial fibrillation: a Mendelian randomization study. *Eur Heart J*. 2019 Apr 21;40(16):1277-1282. PMID: 30721963 [material support, critical revevision]
90. Moayedi Y, Kozuszko S, **Knowles JW**, Chih S, Oro G, Lee R, Fearon WF, Ross HJ, Teuteberg JJ, Khush KK. Safety and Efficacy of PCSK9 Inhibitors After Heart Transplantation. *Can J Cardiol*. 2019 Jan;35(1):104.e1-104.e3. doi: 10.1016/j.cjca.2018.11.004. Epub 2018 Nov 14. PMID: 30595172 [critical revision]
91. Banda JM, Sarraju A, Abbasi F, Parizo J, Pariani M, Ison H, Briskin E, Hannah Wand H, Dubois S, Jung K, Myers SA, Rader DJ, Leader JB, Murray MF, Myers KD, Wilemon K, Shah NH, **Knowles JW**. Finding missed cases of familial hypercholesterolemia in health systems using machine learning. *npj Digital Medicine* 2 , 1–8

92. Kim D, Cholankeril G, Kim SH, Abbasi F, **Knowles JW**, Ahmed A. Increasing Mortality Among Patients With Diabetes and Chronic Liver Disease From 2007 to 2017. Clin Gastroenterol Hepatol. 2019 Jun 17. pii: S1542-3565(19)30649-4. PMID: 31220638 [critical revison]
93. Duell PB, Gidding SS, Andersen RL, Knickelbine T, Anderson L, Gianos E, Shrader P, Kindt I, O'Brien EC, McCann D, Hemphill LC, Ahmed CD, Martin SS, Larry JA, Ahmad ZS, Kullo IJ, Underberg JA, Guyton J, Thompson P, Wilemon K, Roe MT, Rader DJ, Cuchel M, Linton MF, Shapiro MD, Moriarty PM, **Knowles JW**. Longitudinal Low Density Lipoprotein Cholesterol Goal Achievement and Cardiovascular Outcomes Among Adult Patients With Familial Hypercholestrolemia: The CASCADE FH Registry. Atherosclerosis.2019 Oct; 289:85-93. Doi: 10.1016/j.atherosclerosis.2019.08.007. Epub 2019 Aug 19.
94. Cook NL, Pjanic M, Emmerich AG, Rao AS, Hetty S, **Knowles JW**, Quertermous T, Castillejo-López C, Ingelsson E. CRISPR-Cas9-mediated knockout of SPRY2 in human hepatocytes leads to increased glucose uptake and lipid droplet accumulation. *BMC Endocr Disord*. 2019;19(1):115. Published 2019 Oct 29. doi:10.1186/s12902-019-0442-8. PMID: 31664995 [critical revision, material support]
95. Shahbazi M, Cundiff P, Zhou W, Lee P, Patel A, D'Souza SL, Abbasi F, Quertermous T, **Knowles JW**. The role of insulin as a key regulator of seeding, proliferation, and mRNA transcription of human pluripotent stem cells. *Stem Cell Res Ther*. 2019;10(1):228. Published 2019 Jul 29. doi:10.1186/s13287-019-1319-5. PMID: 31358052
96. Fathzadeh M, Li J, Rao A, Cook N, Chennamsetty I, Seldin M, Zhou X, Sangwung P, Gloudemans MJ, Keller M, Attie A, Yang J, Wabitsch M, Carcamo-Orive I, Tada Y, Lusis AJ, Shin MK, Molony CM, McLaughlin T, Reaven G, Montgomery SB, Reilly D, Quertermous T, Ingelsson E, **Knowles JW**. FAM13A affects body fat distribution and adipocyte function. *Nat Commun*. 2020;11(1):1465. Published 2020 Mar 19. doi:10.1038/s41467-020-15291-z. PMID: 32193374
97. Poon AK, Meyer ML, Tanaka H, Selvin E, Pankow J, Zeng D, Loehr L, **Knowles JW**, Rosamond W, Heiss G.Association of insulin resistance, from mid-life to late-life, with aortic stiffness in late-life: the Atherosclerosis Risk in Communities Study. Cardiovasc Diabetol. 2020 Jan 28;19(1):11. doi: 10.1186/s12933-020-0986-y.PMID: 31992297 [concept and design, critical revision]
98. Clarke SL, Reaven GM, Leonard D, Barlow CE, Haskell WL, Willis BL, DeFina L, **Knowles JW**, Maron DJ. Cardiorespiratory Fitness, Body Mass Index, and Markers of Insulin Resistance in Apparently Healthy Women and Men [published online ahead of print, 2020 Jan 9]. *Am J Med*. 2020;S0002-9343(20)30009 7.doi:10.1016/j.amjmed.2019.11.031. Online ahead of print. PMID: 31926863.[concept and design, critical revision]
99. Shiffman D, Louie JZ, Devlin JJ, **Knowles JW**, McPhaul MJ. Gaps in Dyslipidemia Care Among Working-Aged Individuals With Employer-Sponsored Health Care. *J Am Heart Assoc*. 2020;9(9):e015807. doi:10.1161/JAHA.119.015807. PMID: 32319337 [concept and design, critical revision]
100. Kanchan K, Iyer K, Yanek LR, Carcamo-Orive I, Taub MA, Malley C, Baldwin K, Becker LC, Broeckel U, Cheng L, Cowan C, D'Antonio M, Frazer KA, Quertermous T, Mostoslavsky G, Murphy G, Rabinovitch M, Rader DJ, Steinberg MH, Topol E, Yang W, **Knowles JW**, Jaquish CE, Ruczinski I, Mathias RA. Genomic integrity of human induced pluripotent stem cells across nine studies in the NHLBI NextGen program. *Stem*

Cell Res. 2020 May 6;46:101803. doi: 10.1016/j.scr.2020.101803. PMID: 32442913 [critical revision, material support]

101. Jakubosky D, Smith EN, D'Antonio M, Jan Bonder M, Young Greenwald WW, D'Antonio-Chronowska A, Matsui H; i2QTL Consortium, Stegle O, Montgomery SB, DeBoever C, Frazer KA. Discovery and quality analysis of a comprehensive set of structural variants and short tandem repeats. *Nat Commun*. 2020 Jun 10;11(1):2928. doi: 10.1038/s41467-020-16481-5.PMID: 32522985 [material support as part of the GENESIPS consortium which contributed to the proejct]
102. Jakubosky D, D'Antonio M, Bonder MJ, Smail C, Donovan MKR, Young Greenwald WW, Matsui H; i2QTL Consortium, D'Antonio-Chronowska A, Stegle O, Smith EN, Montgomery SB, DeBoever C, Frazer KA. Properties of structural variants and short tandem repeats associated with gene expression and complex traits. *Nat Commun*. 2020 Jun 10;11(1):2927. doi: 10.1038/s41467-020-16482-4.PMID: 32522982 [material support as part of the GENESIPS consortium which contributed to the proejct]
103. de Ferranti SD, Shrader P, Linton MF, **Knowles JW**, Hudgins LC, Benuck I, Kindt I, O'Brien EC, Peterson AL, Ahmad ZS, Clauss S, Duell PB, Shapiro MD, Wilemon K, Gidding SS, Neal W. Children with Heterozygous Familial Hypercholesterolemia in the United States: Data from the Cascade Screening for Awareness and Detection-FH Registry. J Pediatr. 2021 Feb;229:70-77. doi: 10.1016/j.jpeds.2020.09.042. Epub 2020 Sep 22. PMID: 32976895 [conception and design, critical revision]
104. Carcamo-Orive I, Henrion MYR, Zhu K, Beckmann ND, Cundiff P, Moein S, Zhang Z, Alamprese M, D'Souza SL, Wabitsch M, Schadt EE, Quertermous T, **Knowles JW*,** Chang R*. Predictive network modeling in human induced pluripotent stem cells identifies key driver genes for insulin responsiveness. PLoS Comput Biol. 2020 Dec 23;16(12):e1008491. doi: 10.1371/journal.pcbi.1008491. eCollection 2020 Dec. PMID: 33362275 Free PMC article. * co-senior authors
105. Bonder MJ, Smail C, Gloudemans MJ, Frésard L, Jakubosky D, D'Antonio M, Li X, Ferraro NM, Carcamo-Orive I, Mirauta B, Seaton DD, Cai N, Vakili D, Horta D, Zhao C, Zastrow DB, Bonner DE; HipSci Consortium; iPSCORE consortium; Undiagnosed Diseases Network; PhLiPS consortium, Wheeler MT, Kilpinen H, **Knowles JW**, Smith EN, Frazer KA, Montgomery SB, Stegle O. Identification of rare and common regulatory variants in pluripotent cells using population-scale transcriptomics. Nat Genet. 2021 Mar;53(3):313-321. doi: 10.1038/s41588-021-00800-7. Epub 2021 Mar 4. PMID: 33664507 [material support, critical revision]
106. Weale ME, Riveros-Mckay F, Selzam S, Seth P, Moore R, Tarran WA, Gradovich E, Giner-Delgado C, Palmer D, Wells D, Saffari A, Sivley RM, Lachapelle AS, Wand H, Clarke SL, **Knowles JW**, O'Sullivan JW, Ashley EA, McVean G, Plagnol V, Donnelly P. Validation of an Integrated Risk Tool, Including Polygenic Risk Score, for Atherosclerotic Cardiovascular Disease in Multiple Ethnicities and Ancestries. Am J Cardiol. 2021 Jun 1;148:157-164. doi: 10.1016/j.amjcard.2021.02.032. Epub 2021 Mar 3. PMID: 33675770 [critical revision]
107. Liu L, Shenoy SP, Jahng JWS, Liu Y, **Knowles JW**, Zhuge Y, Wu JC. Generation of two heterozygous MYBPC3 mutation-carrying human iPSC lines, SCVIi001-A and SCVIi002-A, for modeling hypertrophic cardiomyopathy. Stem Cell Res. 2021 Mar 11;53:102279. doi: 10.1016/j.scr.2021.102279. PMID: 33743363 [material support]

108. Sarraju A, Valencia A, **Knowles JW**, Maron DJ, Rodriguez F. Diverse Racial/Ethnic Group Underrepresentation in High-Impact Cholesterol Treament Trials. Circulation. 2021 Jun 15;143(24):2409-2411. doi: 10.1161/CIRCULATIONAHA.120.050034. Epub 2021 Jun 14. PMID: 34125567 [study design input and critical design]
109. Kim DS, Gloyn AL, **Knowles JW**. Genetics of Type 2 Diabetes: Opportunities for Precision Medicine: JACC Focus Seminar. J Am Coll Cardiol. 2021 Aug 3;78(5):496-512. doi: 10.1016/j.jacc.2021.03.346.PMID: 34325839
110. Katz SF, Rodriguez F, **Knowles JW**. Health disparities in cardiometabolic risk among Black and Hispanic youth in the United States. Am J Prev Cardiol. 2021 Mar 23;6:100175. doi: 10.1016/j.ajpc.2021.100175. eCollection 2021 Jun.PMID: 34327498
111. Jiang Z, Zhao M, Voilquin L, Jung Y, Aikio MA, Sahai T, Dou FY, Roche AM, Carcamo-Orive I, **Knowles JW**, Wabitsch M, Appel EA, Maikawa CL, Camporez JP, Shulman GI, Tsai L, Rosen ED, Gardner CD, Spiegelman BM, Svensson KJ. Isthmin-1 is an adipokine that promotes glucose uptake and improves glucose tolerance and hepatic steatosis. Cell Metab. 2021 Sep 7;33(9):1836-1852.e11. doi: 10.1016/j.cmet.2021.07.010. Epub 2021 Aug 3.PMID: 34348115 [critical revision, experimental assistance]
112. Abbasi F, Lamendola C, Harris CS, Harris V, Tsai MS, Tripathi P, Abbas F, Reaven GM, Reaven PD, Snyder MP, Kim SH, **Knowles JW**. Statins Are Associated With Increased Insulin Resistance and Secretion. Arterioscler Thromb Vasc Biol. 2021
113. Haider N, Lebastchi J, Jayavelu AK, Batista TM, Pan H, Dreyfuss JM, Carcamo-Orive I, **Knowles JW**, Mann M, Kahn CR. Signaling defects associated with insulin resistance in nondiabetic and diabetic individuals and modification by sex. J Clin Invest. 2021 Nov 1;131(21):e151818. doi: 10.1172/JCI151818.PMID: 34506305 [provided critical reagents, experimental design input, critical revision]
114. Balliu B*, Carcamo-Orive I*, Gloudemans MJ, Nachun DC, Durrant MG, Gazal S, Park CY, Knowles DA, Wabitsch M, Quertermous T, **Knowles JW***, Montgomery SB*. An integrated approach to identify environmental modulators of genetic risk factors for complex traits. Am J Hum Genet. 2021 Oct 7;108(10):1866-1879. doi: 10.1016/j.ajhg.2021.08.014. Epub 2021 Sep 27.PMID: 34582792, *co-senior author on paper
115. Pearson K, Ngo S, Ekpo E, Sarraju A, Baird G, **Knowles JW**, Rodriguez F. Online Patient Education Materials Related to Lipoprotein(a): Readability Assessment. J Med Internet Res. 2022 Jan 11;24(1):e31284. doi: 10.2196/31284. PMID: 35014955; PMCID: PMC8790699.
116. Gorzynski JE, Goenka SD, Shafin K, Jensen TD, Fisk DG, Grove ME, Spiteri E, Pesout T, Monlong J, Baid G, Bernstein JA, Ceresnak S, Chang PC, Christle JW, Chubb H, Dalton KP, Dunn K, Garalde DR, Guillory J, **Knowles JW**, Kolesnikov A, Ma M, Moscarello T, Nattestad M, Perez M, Ruzhnikov MRZ, Samadi M, Setia A, Wright C, Wusthoff CJ, Xiong K, Zhu T, Jain M, Sedlazeck FJ, Carroll A, Paten B, Ashley EA. Ultrarapid Nanopore Genome Sequencing in a Critical Care Setting. N Engl J Med. 2022 Feb 17;386(7):700-702. doi: 10.1056/NEJMc2112090. Epub 2022 Jan 12. PMID: 35020984.
117. Chora JR, Iacocca MA, Tichý L, Wand H, Kurtz CL, Zimmermann H, Leon A, Williams M, Humphries SE, Hooper AJ, Trinder M, Brunham LR, Costa Pereira A, Jannes CE, Chen M, Chonis J, Wang J, Kim S, Johnston T, Soucek P, Kramarek M, Leigh SE, Carrié A, Sijbrands EJ, Hegele RA, Freiberger T, **Knowles JW**, Bourbon M; ClinGen Familial

Hypercholesterolemia Expert Panel. The Clinical Genome Resource (ClinGen) Familial Hypercholesterolemia Variant Curation Expert Panel consensus guidelines for LDLR variant classification. Genet Med. 2022 Feb;24(2):293-306. doi: 10.1016/j.gim.2021.09.012. Epub 2021 Nov 30. PMID: 34906454.
118. Marwaha S, **Knowles JW**, Ashley EA. A guide for the diagnosis of rare and undiagnosed disease: beyond the exome. Genome Med. 2022 Feb 28;14(1):23. doi: 10.1186/s13073-022-01026-w. PMID: 35220969; PMCID: PMC8883622.
119. Gloudemans MJ, Balliu B, Nachun D, Schnurr TM, Durrant MG, Ingelsson E, Wabitsch M, Quertermous T, Montgomery SB, **Knowles JW**, Carcamo-Orive I. Integration of genetic colocalizations with physiological and pharmacological perturbations identifies cardiometabolic disease genes. Genome Med. 2022 Mar 15;14(1):31. doi: 10.1186/s13073-022-01036-8. PMID: 35292083; PMCID: PMC8925074.
120. SL Clarke, C Tcheandjieu, AT Hilliard, KM Lee, J Lynch, KM Chang, D Miller, **Knowles JW**, C O'Donnell, PS Tsao, DJ Rader, PW Wilson, YV Sun, JM Gaziano, TL Asimes, VA Million Veteran Program. Coronary Artery Disease Risk of Familial Hypercholesterolemia Genetic Variants Independent of Clinically Observed Longitudinal Cholesterol Exposure. Circulation: Genomic and Precision Medicine. Lippincott Williams & Wilkins. 2022 Apr. 15(2): e003501.
121. Manhas A, Jahng JWS, Vera CD, Shenoy SP, **Knowles JW**, Wu JC. Generation of two iPSC lines from hypertrophic cardiomyopathy patients carrying MYBPC3 and PRKAG2 variants. Stem Cell Res. 2022 May;61:102774. doi: 10.1016/j.scr.2022.102774. Epub 2022 Mar 31. PMID: 35413566.
122. Ison HE, Clarke SL, **Knowles JW**. Familial Hypercholesterolemia. 2014 Jan 2 [updated 2022 Jul 7]. In: Adam MP, Everman DB, Mirzaa GM, Pagon RA, Wallace SE, Bean LJH, Gripp KW, Amemiya A, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993–2022. PMID: 24404629.
123. Schnurr TM, Katz SF, Justesen JM, O'Sullivan JW, Saliba-Gustafsson P, Assimes TL, Carcamo-Orive I, Ahmed A, Ashley EA, Hansen T, **Knowles JW**. Interactions of physical activity, muscular fitness, adiposity, and genetic risk for NAFLD. Hepatol Commun. 2022 Jul;6(7):1516-1526. doi: 10.1002/hep4.1932. Epub 2022 Mar 15. PMID: 35293152; PMCID: PMC9234625.
124. Groselj U, Šikonja J, Mlinarič M, Kotnik P, Battelino T, **Knowles JW**. Statin therapy is not associated with insulin resistance in children and adolescents with familial hypercholesterolaemia. Atherosclerosis Journal. 2022 Aug 1; 355:91. Doi: DOI: https://doi.org/10.1016/j.atherosclerosis.2022.06.500.
125. Groselj U, Sikonja J, Mlinaric M, Kotnik P, Battelino T, **Knowles JW**. Analysis of Insulin Resistance Among Children and Adolescents in Slovenia With Hypercholesterolemia After Treatment With Statins. JAMA Netw Open. 2022 Sep 1;5(9):e2231097. doi: 10.1001/jamanetworkopen.2022.31097. PMID: 36094507; PMCID: PMC9468884.
126. Lewis EF, Beaty C, Boltze J, Breathett K, Clair WK, de Las Fuentes L, Essien UR, Goodell H, Hinson HE, Kershaw KN, **Knowles JW**, Mazimba S, Mujahid M, Okafor HE, Woo Park K, Schultz J. Creation of the American Heart Association Journals' Equity, Diversity, and Inclusion Editorial Board: The Next Step to Achieving the 2024 Impact Goal. Circulation. 2022 Sep 6;146(10):721-723. doi: 10.1161/CIRCULATIONAHA.122.061450. Epub 2022 Jul 18. PMID: 35862071.

127. Rowland CM, Abbasi F, Shiffman D, **Knowles JW**, McPhaul MJ. The relationship between insulin resistance and ion mobility lipoprotein fractions. Am J Prev Cardiol. 2022 Dec 25;13:100457. doi: 10.1016/j.ajpc.2022.100457. PMID: 36619297; PMCID: PMC9816659.
128. Kim D, Alshuwaykh O, Dennis BB, Cholankeril G, **Knowles JW**, Ahmed A. Chronic liver disease-related mortality in diabetes before and during the COVID-19 in the United States. Dig Liver Dis. 2023 Jan;55(1):3-10. doi: 10.1016/j.dld.2022.09.006. Epub 2022 Sep 29. PMID: 36182570; PMCID: PMC9534558.
129. Diane E MacDougall, Mary P McGowan, Catherine D Ahmed, Daniel J Rader, **Knowles JW**, Katherine Wilemon, Kelly D Myers. DIAGNOSIS OF FAMILIAL HYPERCHOLESTEROLEMIA: A WORK IN PROGRESS. American Journal of Preventive Cardiology. 2023 Jan;13:100414. doi: 10.1016/j.ajpc.2022.100414.
130. Cuchel M, Lee PC, Hudgins LC, Duell PB, Ahmad Z, Baum SJ, Linton MF, de Ferranti SD, Ballantyne CM, Larry JA, Hemphill LC, Kindt I, Gidding SS, Martin SS, Moriarty PM, Thompson PP, Underberg JA, Guyton JR, Andersen RL, Whellan DJ, Benuck I, Kane JP, Myers K, Howard W, Staszak D, Jamison A, Card MC, Bourbon M, Chora JR, Rader DJ, **Knowles JW**, Wilemon K, McGowan MP. Contemporary Homozygous Familial Hypercholesterolemia in the United States: Insights From the CASCADE FH Registry. J Am Heart Assoc. 2023 May 2;12(9):e029175. doi: 10.1161/JAHA.122.029175. Epub 2023 Apr 29. PMID: 37119068.
131. Bielczyk-Maczynska E, Sharma D, Blencowe M, Saliba Gustafsson P, Gloudemans MJ, Yang X, Carcamo-Orive I, Wabitsch M, Svensson KJ, Park CY, Quertermous T, **Knowles JW**, Li J. A single-cell CRISPRi platform for characterizing candidate genes relevant to metabolic disorders in human adipocytes. Am J Physiol Cell Physiol. 2023 Jul 24. doi: 10.1152/ajpcell.00148.2023. Epub ahead of print. PMID: 37486064.
132. Zanetti D, Stell L, Gustafsson S, Abbasi F, Tsao PS, **Knowles JW**; RISC Investigators; Zethelius B, Ärnlöv J, Balkau B, Walker M, Lazzeroni LC, Lind L, Petrie JR, Assimes TL. Plasma proteomic signatures of a direct measure of insulin sensitivity in two population cohorts. Diabetologia. 2023 Jun 17. doi: 10.1007/s00125-023-05946-z. Epub ahead of print. PMID: 37329449.
133. Li J, Jin C, Gustafsson S, Rao A, Wabitsch M, Park CY, Quertermous T, **Knowles JW**, Bielczyk-Maczynska E. Single-cell transcriptome dataset of human and mouse in vitro adipogenesis models. Sci Data. 2023 Jun 16;10(1):387. doi: 10.1038/s41597-023-02293-x. PMID: 37328521; PMCID: PMC10275883.

B. Peer reviewed other papers (editorials, commentaries)

1. **Knowles JW**, Maeda N. Genetic modifiers of atherosclerosis in mice. *Arterioscler Thromb Vasc Biol* 2000; 20(11):2336-2345. Review.
2. **Knowles JW**, Elliott AB, Brody J. A case of complete heart block reverting to normal sinus rhythm after treatment for cardiac invasive Burkitt's lymphoma. *Ann Hematol* 2007; 86(9):687-690.
3. **Knowles JW**, Assimes TL, Li J, Quertermous T, Cooke JP. Genetic susceptibility to peripheral arterial disease: a dark corner in vascular biology. *Arterioscler Thromb Vasc Biol* 2007; 27(10):2068-2078. Review.

4. Wheeler MT, Ho M, **Knowles JW**, Pavlovic A, Ashley EA. Pharmacogenetics of heart failure: evidence, opportunities, and challenges for cardiovascular pharmacogenomics. *J Cardiovasc Trans Res* 2008; 1(1):25-36.
5. deGoma EM**, Knowles JW,** Angeli F, Budoff MJ, Rader DJ. The evolution of traditional risk factors. *Cardiol Rev* 2012 May; 20(3):118-129.
6. Pan S, **Knowles JW.** Exploring predisposition and treatment response--the promise of genomics. *Prog Cardiovasc Dis* 2012 Jul-Aug; 55(1):56-63. Review.
7. Rodriguez F, **Knowles JW**. PCSK9 Inhibition: Current Concepts and Lessons from Human Genetics.Curr Atheroscler Rep. 2015 Mar; 17(3):487. doi: 10.1007/s11883-015-0487-8.PMID: 25637042.
8. Parizo J, Sarraju A, **Knowles JW**. Novel Therapies for Familial Hypercholesterolemia. *Curr Treat Options Cardiovasc Med*. 2016 Nov;18(11):64. doi: 10.1007/s11936-016-04862. Review. PMID: 27620638.
9. Sarraju A, Kim SH, **Knowles JW**. Cardiometabolic Effects of Glucagon-Like Peptide-1 Agonists. *Curr Atheroscler Rep*. 2016 Feb;18(2):7. doi: 10.1007/s11883-016-0558-5. Review. PMID: 26782825.
10. Carcamo-Orive I, Huang NF, Quertermous T, **Knowles JW**. Induced Pluripotent Stem Cell-Derived Endothelial Cells in Insulin Resistance and Metabolic Syndrome. *Arterioscler Thromb Vasc Biol*. **2017** Nov;37(11):2038-2042. doi: 10.1161/ATVBAHA.117.309291. Epub **2017** Jul 20. Review. PMID 28729365
11. Priest JR, **Knowles JW**. Standards of Evidence and Mechanistic Inference in Autosomal Recessive Hypercholesterolemia. *Arterioscler Thromb Vasc Biol.* 2016 Aug;36(8):1465-6. doi: 10.1161/ATVBAHA.116.307714. No abstract available. PMID: 27466618
12. **Knowles JW**. Statins in Familial Hypercholesterolemia: Translating Evidence to Action. *J Am Coll Cardiol*. 2016 Jul 19;68(3):261-4. doi: 10.1016/j.jacc.2016.05.024. No abstract available. PMID: 27417003.
13. Rodriguez F, **Knowles JW**. Enough Evidence, Time to Act! *Circulation*. 2016 Jul 5;134(1):20-3. doi: 10.1161/CIRCULATIONAHA.116.023010. No abstract available. PMID: 27358433.
14. Carcamo-Orive I, Huang NF, Quertermous T, **Knowles JW**. Induced Pluripotent Stem Cell-Derived Endothelial Cells in Insulin Resistance and Metabolic Syndrome. *Arterioscler Thromb Vasc Biol*. **2017** Nov;37(11):2038-2042. doi: 10.1161/ATVBAHA.117.309291. Epub **2017** Jul 20. Review. PMID 28729365
15. Kindt I, Mata P, **Knowles JW.** The role of registries and genetic databases in familial Hypercholesterolemia. *Curr Opin Lipidol*. 2017 April 28. doi: 10.1097/MOL.0000000000000398. PMID: 28169870.
16. **Knowles JW**, Rader DJ, Khoury MJ. Cascade Screening for Familial Hypercholesterolemia And the Use of Genetic Testing. *JAMA*. 2017 July 25. 318(4):381-382. doi: 10.1001/jama.2017.8543.
17. **Knowles JW**, Sarraju A. Is ACS in Young Patients a "Canary in the Coal Mine" for Familial Hypercholesterolemia*?*. *J Am Coll Cardiol*. 2017 Oct 3. 70(14):1741-1743. Doi: 10.1016/j.jacc.2017.08.019.
18. Ingelsson E, **Knowles JW**. Levering Human Genetics to Understand the Relation of LDL Cholesterol with Type 2 Diabetes. *Clinical Chemistry*. 17 May 2017. doi:10.1373/clinchem.2016.268565.

19. **Knowles JW**, Ashley EA. Cardiovascular disease: The rise of the genetic risk score. *PLoS Med.* **2018** Mar 30;15(3):e1002546. doi: 10.1371/journal.pmed.1002546. eCollection **2018** Mar. PMID: 29601582
20. Sarraju A, **Knowles JW**. Genetic Testing and Risk Scores: Impact on Familial Hypercholesterolemia. Front Cardiovasc Med. 2019 Jan 29;6:5. doi: 10.3389/fcvm.2019.00005. eCollection 2019. Review. PMID: 30761309
21. Rao AS, **Knowles JW**. Polygenic risk scores in coronary artery disease. Curr Opin Cardiol. 2019 Jul;34(4):435-440. PMID: 30994529
22. Hegele RA, **Knowles JW**, Horton JD. Delisting *STAP1*: The Rise and Fall of a Putative Hypercholesterolemia Gene. *Arterioscler Thromb Vasc Biol*. 2020;40(4):847-849. doi:10.1161/ATVBAHA.120.314006. PMID: 32208993.
23. Sangwung P, Petersen KF, Shulman GI, **Knowles JW**. Mitochondrial Dysfunction, Insulin Resistance, and Potential Genetic Implications. *Endocrinology*. 2020;161(4):bqaa017. doi:10.1210/endocr/bqaa017. PMID: 32060542
24. Balla S, Ekpo EP, Wilemon KA, **Knowles JW**, Rodriguez F. Women Living with Familial Hypercholesterolemia: Challenges and Considerations Surrounding Their Care. *Curr Atheroscler Rep*. 2020 Aug 20;22(10):60. doi: 10.1007/s11883-020-00881-5. PMID: 32816232
25. Wand H, **Knowles JW**, Clarke SL. The need for polygenic score reporting standards in evidence-based practice: lipid genetics use case. *Curr Opin Lipidol*. 2021 Apr 1;32(2):89-95. doi: 10.1097/MOL.0000000000000733. PMID: 33538426

C. Non-peer reviewed journal articles

1. **Knowles JW**, Maeda N. Dyslipidemia and hypertension: twin killers in renal vascular disease. *Am J Kidney Dis* 2001; 37(6):1322-1323.
2. **Knowles JW**, Reaven G. Usual Blood Pressure and New-Onset Diabetes Risk: Evidence From 4.1 Million Adults and a Meta-Analysis. *J Am Coll Cardiol*. 2016 Apr 5;67(13):1656-7. doi: 10.1016/j.jacc.2015.12.065. No abstract available. PMID:27150694.
3. E Bielczyk-Maczynska, M Zhao, PJH Zushin, TM Schnurr, HJ Kim, J Li, P Nallagatla, P Sangwung, C Park, C Cornn, A Stahl, KJ Svensson, **Knowles JW**. G protein-coupled receptor 151 regulates glucose metabolism and hepatic gluconeogenesis. bioRxiv. 2022 Jan 1. Cold Spring Harbor Laboratory. doi: https://doi.org/10.1101/2022.05.25.493489 (preprint)4
4. Zhang P, Munier JJ, Vergnes L, Wiese CB, Link JC, Abbasi F, Ronquillo E, Munoz A, Kuang YL, Liu M, Sanchez G, Oni-Orisan A, Iribarren C, McPhaul MJ, Nomura DK, **Knowles JW**, Krauss RM, Medina MW, Reue K. X chromosome dosage drives statin–induced dysglycemia and mitochondrial dysfunction. BioRxiv. doi: https://doi.org/10.1101/2022.08.29.505759?. 2022 Aug 29 (preprint and has not been certified by peer review)
5. Watts G, Schwabe C, Scott R, Gladding P, Sullivan D, Baker J, Clifton P, Given B, Hamilton J, Melquist S, Chang T, Zhou R, Martin JS, Gaudet D, Goldberg I, **Knowles JW**, Hegele R, Ballantyne C. RNA interference targeting hepatic angiopoietin-like protein 3 results in prolonged reductions in serum triglyceride and non-HDL-cholesterol

concentrations: first human results with ARO-ANG3. Research Square. 2022 Oct 5. DOI: https://doi.org/10.21203/rs.3.rs-2097342/v1. (preprint)

6. Li J, Jin C, Gustafsson S, Rao A, Wabitsch M, Park CY, Quertermous T, Bielczyk-Maczynska E, **Knowles JW**. Single-cell transcriptome dataset of human and mouse in vitro adipogenesis models. bioRxiv [Preprint]. 2023 Mar 29:2023.03.27.534456. doi: 10.1101/2023.03.27.534456. PMID: 37034809; PMCID: PMC10081256. (preprint)

D. Book Chapters

1. **Knowles JW** (2004). Genes that Modify Susceptibility to Atherosclerosis: Targets for Drug Action. In: M. Wilkins (ed.) Cardiovascular Pharmacogenetics. (pp. 79-106) Heidelberg, Germany: Springer-Verlag. ISBN: 978-3-540-40204-6
2. Pan S, Kim B, Mehta NN, **Knowles JW** (2013). Cardiovascular Genetics and Genomics in Clinical Practice. S. Shah and D. Arnett (ed.) New York: Demos Medical Publishing (in preparation)
3. Fathzadeh M, Hein D, **Knowles JW** (2017). The Human arylamine N-acetyltransferase type 2 gene: Genomics and Cardiometabolic Risk.
4. Youngblom E., **Knowles JW**. Familial Hypercholesterolemia. 2014 Jan 2. In: Pagon RA, Adam MP, Bird TD, et al., editors. GeneReviews™ [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2014. Note updated in 2017 with Mitchel Pariani then in 2020 with Hannah Wand and Hannah Ison. http://www.ncbi.nlm.nih.gov/books/NBK174884/
5. Palmisano BT, **Knowles JW** (2023). Polygenic Risk Scores. In: C. Ballantyne (ed.) Clinical Lipidology. Chapter 7. Elsiever eBook ISBN: 9780323882873

E. Books: None

F. Digital Publications: None

G. Abstracts not published in other forms NOTE: I STOPPED KEEPING TRACK OF ABSTRACTS IN 2016 as with group authorship it became challenging

1. **Knowles J**, Li Z, Goy MF. Expression of a cGMP-stimulated phosphodiesterase in the rat gastrointestinal-tract, as detected by *in-situ* hybridization and functional assay. *Gastroenterology* 1994; 106(4 Suppl A):A1032.
2. Enhanced atherosclerosis and kidney dysfunction in eNOS$^{-/-}$Apoe$^{-/-}$ mice are ameliorated by enalapril treatment. National MD/PhD Student Conference. Aspen, CO 2000
3. Enhanced atherosclerosis and kidney dysfunction in eNOS$^{-/-}$Apoe$^{-/-}$ mice are ameliorated by enalapril treatment. Arteriosclerosis, Thrombosis and Vascular Biology (ATVB) Meeting. Denver, CO 2000
4. Natriuretic peptide receptor A knockout mice have an enhanced response to hypertrophic stimuli in the heart. Keystone Meeting. Snowbird, UT 2000
5. Hodgin JB, **Knowles JW**, Smithies O, Maeda N. Endothelial nitric oxide synthase is not required for atheroreception by 17 beta-estradiol in ApoE -/- mice. *Circulation* 2000 Oct; 102(18 Suppl II):II-318.

6. **Knowles JW**, Assimes T, Priest JR, Tabor H, Southwick A, Volcik K, Boerwinkle E, Grove ML, Wang J, Tabibiazar R, Sidney S, Risch N, Iribarren C, Go A, Myers R, Hlatky M, Fortmann S, Quertermous T. Failure to replicate an association of a single nucleotide polymorphism in the oxidized LDL receptor gene (Orl1) with CAD. AHA Western States Affiliate -Young Investigators Forum, San Francisco, CA, Sept. 2006.
7. †Absher D, Assimes TL, Basu A, Fortmann SP, Go AS, Hlatky MA, Iribarren C, **Knowles JW**, Li J, Risch N, Sidney S, Southwick A, Tang H, Myers RM, Quertermous T. Genome-wide association study for early coronary disease and related phenotypes. Cold Spring Harbor Laboratory Clinical Cardiovascular Genomics Meeting, Oct. 2007. † *alpha order*.
8. Hao K, Yang X, **Knowles JW**, Assimes TL, Cordell HJ, Xie W, Weedon MN, Zhong H, Frayling TM, Suver C, WangI-M, Greenawalt DM, Kemp DM, Abbasi F, Reaven G, Ho L-T, Chuang L-M, Sheu WH-H, Shih K-C, Wang W-C, Ferrannini E, Smith U, Häring H, Pedersen O, Hansen T, Paananen J, Zhang B, Zhu J, Keller M, Attie A, Ingelsson E, Kaplan LM, Tsao PS, Lum P, Schadt EE, Laakso M, Walker M, Hsiung A, The RISC Consortium, Quertermous T. Elucidating gene pathways underlying insulin resistance using integrated approaches of genome-wide association study, expression QTLs and pathway analysis. American Society of Human Genetics (ASHG). Montreal, Oct 2007.
9. Witteles R, **Knowles JW**, Perez M, Morris WH, Spettell CM, Brennan TA, Heidenreich PA. Overuse of left ventriculography. *J Am Coll Cardiol* 2011 Apr; 57(14):E1289.
10. **Knowles JW**, Assimes TL, Tsao PS, Natali S, Mari A, Quertermous T, Reaven G, Abbasi F. Measurement of Insulin-mediated Glucose Uptake: Direct Comparison of the Modified Insulin Suppression Test and the Euglycemic, Hyperinsulinemic Clamp. International Congress on Insulin Resistance. Los Angeles, CA, Nov 2011.
11. **Knowles JW**, Hao K, Assimes TL, Zhu J, Zhang B, Cordell HJ, Xie W, Weedon MN, Zhong H, Frayling TM, Suver C, Wang IM, Greenawalt DM, Kemp DM, Abbasi F, Reaven G, Ho LT, Chuang LM, Sheu WHH, Shih KC, Wang WC, Ferrannini E, Smith U, Häring H, Pedersen O, Hansen T, Paananen J, Keller M, Attie A, Ingelsson E, Kaplan LM, Tsao PS, Lum P, Schadt EE, Laakso M, Walker M, Hsiung A; The RISC Consortium, Quertermous T, Yang X. Elucidating Gene Pathways and Key Regulators Underlying Insulin Resistance via Integration of Genome-Wide Association Data, Expression QTLs, and Pathway and Network Analysis NHLBI Symposium: Genomics: Gene Discover and Clinical Applications for Cardiovascular, Lung and Blood Diseases, Bethesda, MD, Sep 2011.
12. Xie W, Weedon M, Wood A, GENESIS Consortium, Frayling TM, Gall W, Walker M. Genetic Variants Associated with Diabetes Related Circulating Metabolite Levels and Their Role in Type 2 Diabetes and Insulin Sensitivity. American Diabetes Association 72nd Scientific Sessions, Philadelphia, PA, Jun 2012. [poster presentation]
13. Fall T, Xie W, Hao K, Ärnlöv J, Abbasi F, Schadt EE, Boran G, Hansen T, Greenawalt D, Nolan JJ, Pedersen O, Häring H, Ferrannini E, Syvänen AC, Quertermous T, Smith U, Assimes TL, Laakso M, Walker M, **Knowles JW**, Weedon MN, Ingelsson E, Frayling TM, on behalf of the GENESIS Consortium. Mendelian randomisation studies do not support a causal effect of plasma lipids on insulin sensitivity. AHA Scientific Sessions, Los Angeles, CA 2012.
14. **Knowles JW**, Assimes TL, Kiernan M, Pavlovic A, Goldstein BA, Yank V, McConnell MV, Absher D, Bustamante C, Ashley EA, Ioannidis JPA. Pilot randomized trial of

personalized genomics in preventive cardiology. Oral presentation. AHA Scientific Sessions, Los Angeles, CA 2012.

15. Dimas AS, Lagou V, **Knowles JW**, Mägi R, Barker A, Hivert MF, Benazzo A, Rybin D, Assimes T, Quertermous T, Walker M, Barroso I, Langenberg C, Dupuis J, Watanabe RM, Florez JC, McCarthy M, Ingelsson E, Prokopenko I, on behalf of GENESIS, DIAGRAM, and MAGIC. Impact of variation in type 2 diabetes susceptibility loci on physiologic glycaemic traits in non-diabetic individuals. Oral Presentation. AHA Scientific Sessions, Los Angeles, CA 2012.
16. Ivan Carcamo-Orive, Paige Cundiff, Sunita D'Souza, Hope Lancero, Qi Huang, Caroline Hendry, Ana Sevilla, Angela Bayot, Fahim Abbasi, Gerald M. Reaven, Eric Schadt, Ihor Lemischka, **Joshua W. Knowles**, Thomas Quertermous. Modeling insulin resistance through iPSC technology. Abstracts at the 11th International Society for Stem Cell Research Annual Meeting, June 12-15, Boston, MA
17. Paige Cundiff, Sunita D'Souza, Caroline Hendry, Ivan Carcamo-Orive, Ana Sevilla, Hope Lancero, Fahim Abbasi, Angela Bayot, Gaurav Pandey, Thomas Quertermous, Josh W. Knowles, Eric Schadt, Ihor R. Lemischka. The GENESiPS Study: Identifying the Genetics of Insulin Resistance. Abstracts at the 11th International Society for Stem Cell Research Annual Meeting, June 12-15, Boston, MA.
18. Indumathi Chennamsetty, Qi Huang, Ivan Carcamo-Orive, Hope Lancero, Keller MP, Attie AD, Thomas Quertermous, **Joshua W. Knowles**. *Nat1* deficiency impairs mitochondrial function and promotes insulin resistance Insulin Resistance, Diabetes, Cardiovascular Disease World Congress, November 2013, Los Angeles, CA.
19. Zahid Ahmad, MD, Connie Newman, Emily O'Brien, Peter Shrader, Emil M deGoma, Catherine D Ahmed, Patrick M Moriarty, MD, Mac Rae F Linton, Michael D Shapiro, Paul Barton Duell, Christie M Ballantyne, William A Neal, Danielle Duffy, Lisa Hudgins, Linda C Hemphill, James A Underberg, Karol E Watson, Samuel S Gidding, Seth Baum, Katherine Wilemon, Dave Pickhardt, Iris Kindt, Daniel J Rader, Matthew Roe, **Joshua W Knowles.** Diagnosing Familial Hypercholesterolemia (FH) in the United States: Results from the Cascade FH Patient Registry. Oral Presentation. The Endocrine Society's 97th Annual Meeting, San Diego, CA 2015.
20. Emily C. O'Brien, Emil M. deGoma, Patrick M. Moriarty, MacRae F. Linton, Michael D. Shapiro, P. Barton Duell, Christie M. Ballantyne, William A. Neal, Zahid S. Ahmad, Danielle Duffy, Lisa C. Hudgins, Linda C. Hemphill, James A. Underberg, Karol E. Watson, Samuel S. Gidding, Seth J. Baum, Katherine Wilemon, Dave Pickhardt, Iris Kindt, Daniel J. Rader, Matthew T. Roe, **Joshua W. Knowles**. Initial Results from the CASCADE-FH Registry: CAscade SCreening for Awareness and Detection of Familial Hypercholesterolemia. Abstract at the American College of Cardiology 64th Annual Scientific Sessions, San Diego, CA 2015.
21. Payal Kohli, David Waters, Rana Fayyad, Rachel Laskey, David DeMicco, **Joshua W. Knowles**, Gerald Reaven. Incidence of New-Onset Diabetes on Statin Therapy Increases with Increasing Baseline Triglycerides: Data from TNT. Abstract at the American College of Cardiology 64th Annual Scientific Sessions, San Diego, CA, 2015.
22. Indumathi Chennamsetty, Qi Huang, Ivan Carcamo-Orive, Thomas Quertermous, **Joshua W. Knowles**. *Nat1* knockdown results in an insulin resistance phenotype in vitro and in vivo. Finalist for the ACC Young Investigator Award. Abstract at the American College of Cardiology 64th Annual Scientific Sessions, San Diego, CA, 2015.

23. Payal Kohli, **Joshua W. Knowles**, Rana Fayyad, Gerald M. Reaven, Benoit Arsenault, Pierre Amarenco, Rachel Laskey, David Waters. TG/HDL Ratio Predicts Incident Diabetes and CV Events in Prediabetics and Non-Prediabetics in TNT and SPARCL. Abstract at the European Society of Cardiology, London 2015.
24. E.M. deGoma, Z.S. Ahmad, E. O'Brien, I. Kindt, P. Shrader, C.B. Newman, Y. Pokharel, D.J. Rader, P.M. Moriarty, S.S. Gidding, P. Duell, M.D. Shapiro, M.F. Linton, C.M. Ballantyne, D. Duffy, S.J. Baum, L.C. Hemphill, W.A. Neal, L.C. Hudgins, C.D. Ahmed, K.A. Wilemon, M.T. Row, **J.W. Knowles**. LDL-C Levels and Treatment patterns Among Adults with Heterozygous Familial Hypercholesterolemia in the United States: Data from the CASCADE-FH Registry. Abstract at the American Heart Association Scientific Session, Orlando, FL 2015.
25. Colleen Caleshu, Nadine Kasparian, Katherine Edwards, Laura Yeates, Christopher Semsarian, Marco Perez, Euan Ashley, **Joshua Knowles**, Allysonne Smith, Jodie Ingles. An Interdisciplinary Approach to Psychosocial Care for Families with Inherited Cardiovascular Disease. Abstract at the American Heart Association Scientific Session, Orlando FL 2015.
26. I. Chennamsetty, M. Coronado, K. Contrepois, M. Keller, I. Carcamo-Orive, J. Sandin, G. Fajardo, G. Reaven, A. Attie, D. Bernstein, T. Quertermous, **J. Knowles**. Nat1 deficiency is associated with mitochondrial dysfunction and exercise intolerance in mice. Abstract at the Deuel Conference on Lipids, Napa Valley, CA 2016.

VI. Research Grants

A: Current Funding

9/13/19-5/31/24 NIH R01 DK120565, SPO 137047
CRISPR-Based Gene Perturbation Followed By High-Throughput Phenotyping In Cell & Mouse Models To Characterize Novel Insulin Resistance Genes
PI: Knowles (from co-PI status with Erik Ingelsson)

9/1/19-7/31/24 NIH R01DK116750, SPO 130829
Mechanisms of NAT2 regulation of insulin resistance and mitochondrial function
The goal is to use in vitro and in vivo models to characterize a novel insulin resistance gene
PI: Knowles

7/1/19-6/30/23 AHA SPO 141107
Discovery and Characterization of Novel Genes Associated with Risk for Non-Alcoholic Fatty Liver Disease
The goal is to find novel NAFLD genes
PI: Knowles (changed from co-I to PI from Erik Ingelsson)

7/1/18-6/30/23 NIH SPO 128456
PCSK9 Inhibition after Heart Transplantation

The goal of this work is to determine if PCSK9 inhibitors used after heart transplantation will prevent transplant vasculopathy.
Co-Investigator PI: William Fearon

4/01/18-3/31/23 NIH 1R01DK11418301A1
Proteomic determinants of direct measures of insulin resistance
The goal of this work is to identify proteins and genetic determinants of proteins that are associated with insulin resistance
Co-Investigator PI: Tim Assimes

8/1/23-7/31/27 NIH P30DK116074
Stanford Diabetes Research Center (SDRC)
I am the co-leader of the "Metabolism" working group within the SDRC.

B. Research Grants (*pending*)

9/1/23-11/30/25 NIH 1R01 DK137889-01, SPO 305952
Molecular Mechanisms of Insulin Resistance Associated Loci
Priority score 7th%
PI: Knowles

C. Research Grants (*past*)

08/1/19-7/31/21 US-ISRAEL BSF SPO 134388
Bilateral Science Foundation
An effort to identify FH patients in the databases of Clalit Health Services using machine learning
PI: Knowles

2/1/19-1/31/21 ADA #1-19-JDF-108, SPO 136403
Mechanisms of insulin resistance caused by human NAT2 deficiency
The goal of this work is to understand the underlying basis of the effect of NAT2 on insulin resistance and mitochondrial dysfunction
PI: Knowles

2/20/18-1/31/19 AHA/Brigham and Women's Hospital
Early detection of coronary artery disease in familial hypercholesterolemia
The goal of this work, part of the AHA "One Brave Idea" project is to perform deep phenotyping on patients with familial hypercholesterolemia
Co-Investigator PI: Euan Ashley

8/1/17-7/31/21 NIH U41HG009649, SPO 126378
Clinical Genome Resource (ClinGen)
Major goal is to create a unified, public, and freely available database of genetic alterations relevant to clinical care. Our ultimate goal is to empower clinicians, genetic counselors, and patients to make informed decisions based

on DNA testing. I am a co-Investigator of the ClinGen grant and the co-leader of the group focused on Familial Hypercholesterolemia.
Co-Investigator PI: Carlos Bustamante, Tom Montine

9/01/16-6/30/20 NIH 1R01DK106236,SPO 118336
Beyond GWAS of insulin resistance: an integrated approach to translate genetic association to function
The goals of this work are to use computational, in vitro and zebrafish models to uncover the function of insulin resistance GWAS variants.
PI: Knowles (changed from co-I to PI from Erik Ingelsson

7/01/16-6/30/19 Doris Duke Charitable Trust SPO#: 124038
Statin-associated diabetes: Identifying risk factors and physiologic mechanisms
The goal is to understand the mechanism for statin-induced diabetes. We will perform a small clinical trial to assess insulin sensitivity and insulin secretion with gold standard measures before and after statin therapy.
PI: Knowles

04/1/16-3/31/20 NIH R01DK107437-01, SPO 119387
Molecular Mechanisms of Insulin Resistance Associated Loci
The goals of this work are to identify causal variation and causal genes in type 2 diabetes GWAS loci and to initiate studies to determine the mechanism of the association.
PI: Knowles

01/1/16-12/31/18 Merck SPO # 123020
Evaluation of FAM13A as in insulin resistance gene
Co-Investigator PI: Thomas Quertermous

8/1/15-7/31/20 NIH R01 HL12866605
Escalating proportion of weight loss maintainers prior to weight loss
I adjudicate adverse outcomes during this clinical weight loss trial
Co-Investigator PI: Michaela Kiernan

01/1/15-12/31/17 Amgen SPO #118131
FIND FH: A STRIDE based approach for identification of FH
PI: Knowles

01/1/15-12/31/16 American Heart Association SPO # 116769/15IRG222930034
Summer 2014 Innovative Research Grant
Use of electronic phenotyping and machine learning to identify familial hypercholesterolemia in EHRs
PI: Knowles

1/01/15-12/31/15 Stanford 2014 CVI Seed Grant

Exploring the Role of Maternal Insulin Resistance in Congenital Heart Defects
PI: Knowles

12/20/12-1/7/14 Stanford 2012 CVI Seed Grant
Hybrid genotyping of a well-phenotyped healthy control population as a community resource for exome studies
Co-PI Knowles, w/Jason Merker, MD, PhD (Genetics)

10/1/11-9/30/12 Stanford CVI Seed Grant 1091650-310-GHAKD
A pilot randomized trial of personalized genetics in preventive cardiology
PI: Knowles

07/1/11-6/30/16 NIH 1 U01 HL107388-01
Next Generation Genetic Association Studies (RFA HL11-006)
Identifying the gene networks of insulin resistance: the GENESIPS study
Co-Investigator PI: Thomas Quertermous

10/1/10-9/30/15 Leducq Foundation Network Grant
Molecular mechanisms of novel genes associated with plasma lipids and cardiovascular disease
Contributor PI: Thomas Quertermous
Overall PI: Dan Rader (Penn)

7/1/10-6/30/15 American Heart Association 10FTF3360005 SPO # 48273
National Fellow to Faculty Award
Identification and characterization of genetic determinants of insulin resistance
PI: Knowles

5/1/10-4/30/11 Stanford CTSA Seed Grant, Innovation Award in Population Science
Supported by NIH/NCRR CTSA UL1 RR025744
A pilot randomized trial of personalized genetics in preventive cardiology
PI: Knowles

6/5/08-6/4/09 Rosetta Inpharmatics LLC
Genome-wide association study for determinants of insulin sensitivity
Co-Investigator PI: Thomas Quertermous, MD

4/1/08-4/1/09 Cardiovascular Institute Seed Grant, 2008
Genome-wide association study for determinants of insulin sensitivity
Co-Investigator PI: Thomas Quertermous, MD

9/25/06-7/31/09 NIH R01 HL087647
Whole genome association for early coronary artery disease and related phenotypes

Co-Investigator PI: Thomas Quertermous, MD

9/1/06-9/1/08 American Heart Association 0625154Y
Postdoctoral Grant
Replication and functional studies of a polymorphism in the LOX-1 gene that alters susceptibility to coronary disease
PI: Knowles

9/1/06-9/1/07 LCIC Grant SPO # 37759
Private Foundation Fellowship Grant
Replication and functional characterization of a polymorphism in the LOX-1 gene that alters susceptibility to coronary disease
PI: Knowles

4/1/06-4/1/07 Cardiovascular Institute Seed Grant CVI Award EAFGS
Genetic determinants of coronary artery disease
Co-Investigator PI: Thomas Quertermous, MD

1/1/06-1/1/07 Dean's Post-Doctoral Fellowship Award, Stanford
Identification of genetic polymorphisms that alter susceptibility to coronary artery disease
PI: Knowles

10/1/03-9/31/08 NIH RO1 HL075774
Genetic determinants of peripheral arterial disease
Co-Investigator. co-PI: John Cooke, Thomas Quertermous, MD

VII. Clinical Trials

Current

2018-2025 **Site PI for clinical trials for Inclisiran (The Medicines Co -> Novartis):**
ORION-8, (PI: Joshua Knowles - SPO 150227) Medicines Company
A long term extension trial of the Phase III lipid-lowering trials to assess the effect of long term dosing of inclisiran given as subcutaneous injections in subjects with high cardiovascular risk and elevated LDL-C

2018-2022 **Site co-I for clinical trial for Lp(a) lowering trial (Akcea)**
AKCEA ISIS 681257-CS6 (PI Abha Khandelwal - SPO 127507)
A Randomized, Double-blind, Placebo-Controlled, Dose-Ranging Phase 2 Study of ISIS 681257 Administered Subcutaneously to Patients with Hyperlipoproteinemia(a) and Established Cardiovascular Disease (CVD)

Past

A Pilot Randomized Trial of Personal Genomics for Preventive Cardiology
Role: Stanford site Principal Investigator SPO# 40262
Type: Investigator initiated, single site

Sponsor: Stanford start – stop dates: 10/1/10-5/1/13
Subjects: Enrolled 100 of a planned 100 patients at Stanford
Findings published
NCT ID: **NCT 00248079**

Relationship Between Insulin Resistance and Statin Induced Type 2 Diabetes, and Integrative Personal Omics Profiling
Role: Stanford site Principal Investigator SPO# 124038
Type: Investigator initiated, single site
Sponsor: Stanford start – stop dates: 4/24/15-2/1/20
Subjects: Enrolled 75 patients, paper under review
NCT ID: **NCT 02437084**

Pending

VIII. Patents: None

IX. Editoral Service

A. Editorial Board Memberships

2017- Associate Editor, *Circulation: Genomic and Precision Medicine*
2019- Editoral Board, *American Journal of Preventive Cardiology*
2022- Editorial Board Member, *AHA Journal Equity, Diversity, and Inclusion*

B. Other Peer Review Activities

Reviewer: Scientific Journals
2008-pres. *Cardiovascular Drugs and Therapy, Diabetes*
2008-pres. *Circulation Research*
2008-pres. *Journal of Molecular and Cellular Cardiology*
2011-pres. *Diabetes Care, Diabetologica*
2012-pres. *Circulation: Cardiovascular Genetics, ATVB, European Journal of Clinical Investigation*
2013-pres. *BMC-Genetics, Genomics in Medicine, Circulation*
2014-pres *Diabetes, American Heart Journal*
2015-pres *PLoS One, PLoS Genetics*
2016-pres *JAMA, Science*
2009-pres. *Circulation*
2009-pres. *New England Journal of Medicine*
2011-pres. *Journal of Clinical Investigation*
2019-pres *PNAS*
2020-pres *Nature, Genome Medicine*

Books
2016 Assistant Editor, Hurst's the Heart. 50th year/fourteenth edition
Chief Editors: Robert Harrington, Valentin Fuster

X. Service as Grant Reviewer

2018- Reviewer for the Maternal Child Heath Research Initiative, Stanford (LPCH)
2019- Reviewer: Doris Duke Foundation
2019- Reviewer: ADA
2019- Reviewer: Burroughs Welcome Trust
2020- Reviewer: The Thresher Foundation
2020- Reviewer: US Department of Defense
2020- Reviewer, Norwegian Ministry of Science
2020- Reviewer, Bergen University, Norway
2021- Reviewer, Snow Medical Research Foundation, Australia

XI. University Administrative Services

A. Committee service

2008-2009	Stanford Initiative in Human Genetics and Genomics, Organizer, campus-wide seminar series on human genetics topics
2013-2015	Stanford Cardiovascular Institute (CVI): Steering Committee
2014-current	Stanford Grant Writing Academy,
2015-2017	Co-director (with David Maron), inpatient General Cardiology Service
2015-2019	Data Safety Monitoring Board, AF Clinical Trial (PI: Dr. Narayan)
2017-current	Stanford Diabetes Center Research Symposium: Organizing committee annually Prof Seung Kim, director)
2019-current	DSMB, Effect of dietary high fiber on personal 'omics (PI: Dr. Snyder)
2019-current	Stanford/UPenn Cardiovascular Symposium (2019): Prof Erik Ingelsson chair (hosted by the CVI led by Prof. Joe Wu)
2020-current	Stanford Committee on Residency and Fellowship Interviews post COVID (led by Prof Abraham Verghese)
2021-2022	Biomedical Informatics Faculty Search Committee, Chair: Nigam Shah

B. Leadership roles

7/2018-Present	Program Director, Stanford Cardiovascular Medicine Fellowship program
7/2017-Present	Co-director (with Joy Wu and Vinicio de Jesus Perez, Paul Bollyky), Stanford Translational Investigator Pathway for Internal Medicine Residents
7/2017-Present	Co-leader, section on Metabolism and Signaling, Stanford Diabetes Research Center (Seung Kim, PI)
7/2017- 7/2018	Associate Program Director, Stanford Cardiovascular Medicine Fellowship program
7/2014-Present	Director, Familial Hypercholesterolemia Clinic, Stanford Center for Inherited Cardiovascular Disease

Stanford Conference Organizing Committees

- CARDIA Conference, Stanford CME course (Dr. Euan Ashley, director): Organizing committee (2014, Monterey; 2016, Napa; 2018, Stanford)

- Stanford/Amgen meeting on Genetics and Drug Discovery: Co-director with Dr. Joe Wu, (2017)

XI. Service to Professional Organizations

A. Membership

9/2012-current Fellow, American College of Cardiology
11/2012-current Fellow, American Heart Association
8/2019-current Fellow, National Lipid Association
1/2021-current Fellow, American Society for Preventive Cardiology

B. Committee Service

International committees and task forces:

2013-2017 International advisory board: Canadian Familial Hypercholesterolemia (FH) registry. Canadian effort led by Dr. Jacques Genest, McGill University
2013-2015 International advisory board: International FH Foundation
Chair: Gerald Watts, Australia

National committees and task forces:

2013- Organizing Committee and co-Host
The Family Heart Foundation Annual Summit: annual event
2013-2015 Writing Group, American Heart Association
AHA Scientific Statement on Familial Hypercholesterolemia
Chair of writing group: Sam Gidding
2013-2015 Member, Clinical Lipidology, Lipoprotein Metabolism & Thrombosis Committee of the Arteriosclerosis, Thrombosis and Vascular Biology (ATVB) Council & Lifestyle and Cardiometabolic Health (formerly NPAM) Council for the term dates, July 1, 2013 - June 30, 2015,
Chair of Council: Eliot Brinton

Local committees and task forces
2011 Pacific Lipid Association (part of the National Lipid Association) Familial Hypercholesterolemia (FH) working group

C. Leadership Roles

2012-pres. FH Foundation (Patient-led, 501c3 advocacy group/charitable organization)
Chief Medical or Chief Research Advisor
The goal of this organization is to improve the diagnosis and treatment of FH and raise awareness through information and advocacy.
Major initiatives include: Designing and implementing a National FH Patient Registry in collaboration with the Duke Clinical Research Institute (CASCADE FH); Participating in the AHA Scientific Statement on FH; Testifying at the FDA in favor of new therapies for homozygous FH (November, 2012) and heterozygous FH (Summer, 2014); Serving as a member of the FH Foundation Board of Directors; Overseeing the scientific

content of informational materials; Collaborating with the Scientific Advisory Board of the FH Foundation and other interested groups such as the AHA, National Association of Genetic Counselors, National Lipid Association, Preventive Cardiology Nurses Association. Writing application for a new, FH-specific ICD10 code for FH, which became effective 10/1/16. Pioneering "big data" approaches to identify FH patients in EMRs (FIND FH initiative)(published in 2019).

National/International Conference Organizing Committees

- FH Foundation International FH Summit: Organizing committee and co-Host (2013, Annapolis; 2014, New York; 2015; Pasadena, 2016; Dallas; 2017, Miami; 2018, Los Angeles; 2019, Atlanta; 2020 virtual, Washington DC, 2023
- ACC "Think Tank" on lipids, Washington 2017: Organizing committee

XIII. Presentations

A. Invited presentations

1. *Genome Wide Association Studies of Cardiovascular Disease*
   Stanford Biostatistics Seminar
   Stanford, CA January, 2008
2. *The Athlete's Heart*
   Senior Games of California
   Stanford, CA April, 2010
3. *Genetics of Cardiovascular Disease*
   Stanford School of Medicine, Introductory Epidemiology Course [Instructor: Rita Popat]
   Stanford, CA March, 2010
4. Panelist, Speed Science
   Stanford Cardiovascular Institute Member Retreat
   Stanford, CA September, 2010
5. *Genome wide association study for determinants of insulin sensitivity*
   Vanderbilt University, Cardiology Special Seminar
   Nashville, TN December, 2010
6. *Genome wide association study for determinants of insulin sensitivity*
   University of Pennsylvania, Cardiology Special Seminar
   Philadelphia, PA December, 2010
7. *Personalized medicine and genomics*
   Stanford School of Medicine, Introductory Epidemiology Course *(GENE210)*
   Stanford, CA April, 2011
8. *A pilot randomized trial of personal genomics for preventive cardiology*
   Stanford Medical School Lecture *(MED223)*
   Stanford, CA June, 2011
9. *Genetics of Cardiovascular Disease*
   Stanford SIMR Summer Research Program
   Stanford, CA June, 2011
10. *Atherogenesis*

Stanford EXPLORE Summer Research Program
Stanford, CA June, 2011

11. *Health and Wellness for the Busy Executive*
Stanford Executive Program, Stanford Graduate School of Business
Stanford, CA June, 2011
12. *The Heart*
Stanford School of Medicine, Lecture for 1st year medical students
Stanford, CA July, 2011
13. *Atherogenesis*
Stanford Cardiology Fellow Educational Lecture Series
Stanford, CA Fall, 2011
14. *Genetics of Cardiovascular Disease*
CME talk for Stanford Internal Medicine group
Stanford, CA July, 2011
15. *Elucidating Gene Pathways and Key Regulators Underlying Insulin Resistance via Integration of Genome-Wide Association Data, Expression QTLs, and Pathway and Network Analysis*
NHLBI Symposium: Genomics: Gene Discover and Clinical Applications for Cardiovascular, Lung and Blood Diseases
Bethesda, MD September, 2011
16. *Impacts of Genomics on Disease Treatment*
SAMMS Panel Discussion (School of Medicine initiative to foster cross disciplinary collaboration)
Stanford, CA December, 2011
17. *Randomized trial of personal genomics in preventive cardiology*
Stanford Cardiology Grand Rounds
Stanford, CA January, 2012
18. *Genetics of Cardiovascular Disease*
Stanford School of Medicine, Introductory Epidemiology Course [Instructor: Rita Popat]
Stanford, CA February, 2012
19. *Cardiovascular Genetics*
Apple Computers, Health Series
Cupertino, CA March, 2012
20. *A randomized trial of personal genomics for preventive cardiology: design and challenges*
American Heart Association National Meeting
Los Angeles, CA November, 2012
21. *Genetics of Cardiovascular Disease*
Stanford SIMR Summer Research Program
Stanford, CA June, 2012
22. *Alcohol ablation or myectomy: Which for Whom* (Panel discussion)
CARDIA (Cardiac arrhythmias, sudden death, inherited cardiovascular disease, athletes
Joshua W. Knowles, David Lee, Iocopo Olivoto, Martin Maron
Monterey, CA June 2012
23. *Focus on FH*
Stanford Cardiology Grand Rounds
Stanford, CA September, 2012

24. *Focus on FH*
    Kaiser Permanente San Francisco
    San Francisco, CA October, 2012
25. *Focus on FH*
    Stanford Internal Medicine Noon Conference
    Stanford, CA October, 2012
26. *Approach to the Statin Intolerant Patient*
    Stanford CME program
    Stanford, CA October, 2012
27. *Focus on FH*
    Medical Genetics Grand Rounds, Stanford
    Stanford, CA November, 2012
28. *Cardiovascular Genetics*
    Stanford School of Medicine, Introductory Epidemiology Course [Instructor: Rita Popat]
    Stanford, CA February, 2013
29. *FH Roundtable, hosted by the National Lipid Association*
    Discussion of the role of the FH Foundation in the future of FH care in the US
    New Orleans, LA February, 2013
30. *Approach to the Statin Intolerant Patient*
    Stanford Cardiology Grand Rounds, Stanford
    Stanford, CA March, 2013
31. *Pre-hypertension and hypertension in the young adult*
    Stanford Vaden Health, CME, Stanford
    Stanford, CA April, 2013
32. *FH: Genetics, screening and treatment*
    Preventive Cardiology Nurses Association National Meeting
    Las Vegas, NV May, 2013
33. *FH: What we don't know can hurt us!*
    Why we can't wait: Conference to eliminate health disparities in genomic medicine
    San Francisco, CA May, 2013
34. *Approach to the Statin Intolerant Patient*
    Stanford Internal Medicine Resident lecture series, Stanford and Palo Alto VA
    Stanford, CA August, 2013
35. *What is it like to be a cardiologist?*
    Stanford Explore Series (High school students)
    Stanford, CA August, 2013
36. *Nuts and Bolts of Genetic Testing for FH*
    National Lipid Association Regional Meeting
    Baltimore, MD September, 2013
37. *CASCADE FH registry: Announcing the launch of the national patient registry for FH*
    FH Foundation's First Annual National FH Summit
    Annapolis, MD September, 2013
38. *AHA/ACC 2013 Guidelines on Lipids*
    Stanford Cardiology Fellows Talk
    Stanford , CA December, 2013
39. *Insulin resistance: Genes and Model Systems*

Stanford Endocrinology Grand Rounds
Stanford , CA January, 2014

40. *AHA/ACC 2013 Guidelines on Lipids*
Stanford Internal Medicine
Stanford , CA January, 2014
41. *AHA/ACC 2013 Guidelines on Lipids*
Stanford Medical Students
Stanford , CA January, 2014
42. *Lipoprotein (a): An emerging causal risk factor for CVD*
Stanford Cardiology Grand Rounds, Stanford
Stanford, CA March 2014
43. *iPSCs as model systems for insulin resistance*
Cardiovascular and Pulmonary Breakout Session, Stanford
Stanford, CA April 2014
44. *New Lipid Guidelines*
8th Annual Cardiology for the Primary Care Practitioner
Stanford, CA October, 2014
45. *FH: Challenges and Opportunities*
Regeneron Familial Hypercholesterolemia Symposium
Tarrytown, NY December, 2014
46. *How We Can (and will) Use Genetics to Improve Cardiac Health*
Stanford Heart Fair
Palo Alto, CA February, 2015
47. *SDSI Flagship Project Presentation: "Use of electronic phenotyping and machine learning algorithms to identify familial hypercholesterolemia patients in electronic health records"*
Data Science Workshop
Stanford, CA April, 2015
48. *Identification* and characterization of NAT2 as an insulin resistance gene
CEHG Genetics and Society Symposium 2015
Stanford, CA April, 2015
49. *Modeling insulin resistance using induced pluripotent stem cells*
12th Stem Cell Summit 2015
Boston, MA April, 2015
50. *Preventive Cardiology: Review of the New Cholesterol Treatment Guidelines, A Farewell to LDL Targets? Really?*
Panel: Joshua Knowles, David Maron, Michael McConnell, Sandra Tsai and Mary Ann Champagne
Stanford Cardiovascular Spring Series
San Jose, CA June, 2015
51. *Familial Hypercholesterolemia: Hidden in Plain Sight*
Stanford Internal Medicine Noon Conference
Stanford, CA August, 2015
52. *MED 223 course: Cardiovascular Research & Medicine Lectures*
Stanford, CA January 2016
53. *Having a healthy heart now and into the future*
The Office of Planned Giving for Stanford Medicine Luncheon
Menlo Park, CA February 2016

54. *Heart Disease Prevention: What You Need to Know*
Stanford Heart Month Community Talk
Stanford, CA February, 2016
55. *FH: The present and future*
Invitae meeting attached to Cardiometabolic Health Congress
San Francisco, CA March 2016
79. *Genetic Variants Underlying Cardiovascular Disease*
The Cooper Institute
Dallas, Texas November 2016
56. *How Heart Doctors Stay Heart Healthy*
Health Matters @ Stanford
Palo Alto, CA May 2017
57. *NAT2 as an insulin resistance gene*
2017 Duke-Stanford Cardiovascular Research Symposium
Durham, North Carolina May 2017
58. *New Lipid Trial—LPA Antisense*
Vascular Surgery Conference
Stanford, CA July 2017
59. *Familial Hypercholesterolemia: The epitome of personalized medicine*
Kaiser San Francisco Cardiology Fellowship Program
San Francisco, CA October 2017
60. Insulin Resistance: Insight from genetic studies
UC Berkeley Seminar
Berkeley, CA November 2017
61. Lipids
Stanford Internal Medicine Resident Ambulatory
Stanford, CA November 2017
62. Lipids
Stanford Internal Medicine Resident Ambulatory
Stanford, CA November 2017
63. *ACHD fellows on management of dyslipidemia including statin therapy and PCKS9 inhibitors*
Stanford, CA January 2018
64. *Familial Hypercholesterolemia: the epitome of personalized medicine*
Stanford MED 223 lecture
Stanford, CA February 2018
65. *Insulin Resistance: Molecular Insights*
GI Symposium
Stanford, CA March 2018
66. *Insulin Resistance: Molecular Insights*
GI Symposium
Stanford, CA March 2018
67. *Familial Hyperlipidemia*
Endocrine Fellows' Teaching Conference
Stanford, CA May 2018
68. *Lipids*
Internal Medicine Resident Ambulatory Lectures

Stanford, CA May 2018

69. *Drugs for arteriosclerosis*, Science of Medicine SOM221 Lecture
    Stanford School of Medicine
    Stanford, CA June 2018
70. Statin-associated diabetes: Identifying risk factors and physiologic mechanisms
    2018 Doris Duke Charitable Foundation
    New York City October 2018
71. Dyslipidemia
    ACHD Lecture
    Stanford, CA October 2018, 2019, 2020, 2021
72. Towards Inclusive Exellence in Cardiology Training Programs
    Association of Black Cardiologists, Inc.
    Virtual Presentation November 2021
73. How we can (and will) use genetics to improve cardiovasculat health
    Division of Cardiovascular Diseases and the Educational Committee at Mayo Clinic Arizona
    Virtual Presentation December 2021
74. Upstander Intervention
    Department of Medicine Leadership retreat, Facilitator
    Virtual Presentation November 2021
75. Lipid Management/Hyperlipidemia
    Stanford FM Residents
    Virtual Presentaton May 2022
76. *Finding Casual Genes for Insulin Resistance*
    Duke-Stanford Cardiovascular Research Symposium
    Durham, NC June 2022
77. *Machine Learning to Identify Undiagnosed FH Patients: A Collaboration Involving FH Foundation*
    Stanford and UPennPenn-Stanford CVI Symposium
    Philadelphia, PA Oct 2022

B. National and Regional Meetings

1. *Enhanced atherosclerosis and kidney dysfunction in eNOS$^{-/-}$Apoe$^{-/-}$ mice are ameliorated by enalapril treatment*.
   National MD/PhD Student Conference
   Aspen, CO 2000
2. *Natriuretic peptide receptor A knockout mice have an enhanced response to hypertrophic stimuli in the heart.*
   Keystone Meeting
   Snowbird, UT 2000
3. *Genome Wide Association Study for Early Onset Coronary Disease and Related Phenotypes*.
   Cold Spring Harbor Meeting on Clinical Cardiovascular Genetics
   Cold Spring Harbor, NY November, 2007
4. *FH: A public Health Perspective*
   American Heart Association National Meeting
   Dallas, TX November, 2013

5. *Genetic and Functional Analyses Identify NAT2 as an Insulin Sensitivity Locus*, presented on behalf of the GENESIS consortium
   American Heart Association National Meeting
   Chicago, IL November, 2014
6. *FIND FH Initiative*
   2014 FH Foundation 2nd Annual International FH Summit
   New York, NY October, 2014
7. *FH: Integrating Genetic Information into Clinical Care*
   American Heart Association National Meeting
   Chicago, IL November, 2014
8. *How genetics can improve cardiometabolic health*
   12th Annual World Congress on Insulin Resistance, Diabetes & CVD
   Los Angeles, CA November, 2014
9. *ACC.15 Young Investigator Awards Competition: ACC Herman Investigator Awards in Molecular and Cellular Cardiology*
   2nd Place: Nat1Knowckdown Results in an Insulin Resistance Phenotype in Vitro and in Vivo
   American College of Cardiology
   San Diego, CA March, 2015
10. *A Patient-Centered Approach to Lipid Management in the Complicated Patient*
    Preventive Cardiology Nurses Association National Meeting
    Anaheim, CA April, 2015
11. *CASCADE FH*
    FH Foundation Global Summit
    Pasadena, CA September, 2015
12. *Advances in Identifying and Treating Familial Hyperlipidemia: What's new in Familial Hypercholesterolemia Guidelines*
    American Heart Association National Meeting
    Orlando, FL November, 2015
13. *Insulin Resistance: It's In Your Genes*
    13th World Congress on Insulin Resistance, Diabetes, and Cardiovascular Disease
    Washington, DC November 2015
14. *FIND FH Initiative: Finding Undiagnosed Familial Hypercholesterolemia Patients in the US*
    Presenters: Joshua Knowles and Kelly Myers
    CMHC West, Cardiometabolic Health Congress
    San Francisco, CA March 2016
15. *Putting it all in Perspective: Panel Discussion and Q&A*
    Discussants: J. Knowles, M. Budoff, S. Fazio, K. Myers, J. Underberg
    CMHC West, Cardiometabolic Health Congress
    San Francisco, CA March 2016
16. *USA Cascade FH One Year Data/Find FH/New Recommendations*
    National Lipid Association Meeting
    New Orleans, LA May 2016
17. *Molecular Diagnosis of FH: An Ever Expanding Spectrum of Mutations*
    2016 ASPC Congress on Atherosclerotic Cardiovascular Disease Prevention
    Boca Raton, FL September 2016
18. *ASPC Congress on ASCVD Prevention: FH Foundation: Clinical Initiatives Update*

2016 Congress on Atherosclerotic Cardiovascular Disease Prevention
Boca Raton, FL September 2016

19. *Landscape of FH: CASCADE FH™*
    2016 FH Global Summit
    Dallas, Texas October 2016
20. *Leveraging Big Data for Precision Health*
    2016 American Heart Association Conference
    New Orleans, Louisiana November 2016
21. *Precision Public Health in Action: Preventing Heart Disease and Cancer from Inherited Mutations*
    NCI Webinar December 2016
22. *Heart Teams in Action: Complex Coronary Revascularization*
    American College of Cardiology 66th Annual Scientific Session & Expo
    Washington, DC March 2017
23. *New Perspectives on CAD Risk Factors*
    American College of Cardiology 66th Annual Scientific Session & Expo
    Washington, DC March 2017
24. *Distinguishing FH from non-FH: Right Therapy, Right Patient, Right Time*
    American College of Cardiology: Address the Risk Think Tank
    Washington, DC September 2017
25. *CASCADE FH Registry: Data on Ethnic and Racial Disparities*
    2017 FH Global Summit
    Miami, Florida September 2017
26. *It's Time for Consensus: Defining Familial Hypercholesterolemia*
    CMHC West Congress
    Las Vegas, CA May 2018
27. *Knowledge to improve Care and Research and Advocacy*
    FH 2018 Global Summit
    Marina del Rey, CA October 2018
28. *Clinical Management of Familial Hypercholesterolemia*
    American Heart Association
    Chicago, IL November 2018
29. *Overview of Cascade Screening and the FH Cascade Registry*
    Reducing the Population Burden of Familial Hypercholesterolemia
    Bethesda, MD November 2018
30. *Understanding How Statins Caused Increased Risk of Diabetes*
    World Congress Insulin Resistance Diabetes and Cardiovascular Disease
    Los Angeles, CA November 2018
31. *New Lipid Guidelines and Therapies*
    Cardiology for the PCP, Stanford CME course
    Stanford, CA May 2019
32. *Machine Learning in Cardiovascular Disease*
    AHA Research Leaders Academy
    Baltimore, MD September 2019
33. *Progress in Familial Hypercholesterolemia over the last 10 years*
    FH Foundation International Summit,

Atlanta, GA October 2019

34. *Polygenic Risk Scores*
    National Lipid Association National Sessions,
    Virtual June 2020
35. *CAC in FH*
    AHA National Sessions
    Virtual Nov 2020
36. *Will Gene Editing Ever Be Used for Lipid Disorders,*
    National Lipids Associateion Meeting
    Phoenix, AZ June 2022
37. *Machine Learning to Identify Undiagnosed FH Patients: A Collaboration Involving FH Foundation, Stanford and UPenn*
    Penn-Stanford CVI Symposium
    Philadelphia, PA Oct 2022

C. International Meetings

1. *How we can (and will) use genetics to improve cardiovascular health*
   International Symposium: Genomics and Personalized Preventive Health
   Génome Québec, Canada February, 2014
2. *CASCADE FH registry*
   Canadian Cardiology Society Meeting, FH registry working group
   Montreal, Canada October, 2013
3. *International Perspectives on FH*
   Canadian Collaborative Research Network: FH Canada
   Vancouver, BC October 2017

D. Visiting Professorships

Duke Medicine:
Genomic and Precision Medicine Forum Presentation *How we can (and will) use genetics to improve Cardiovascular Health*
Durham, NC April 2016

The Ohio State University
Familial Hypercholesterolemia: The Present and the Future
Columbus, OH November 2016

Mass. General Hospital
Familial Hypercholesterolemia
Cardiology Grand Rounds
Boston, MA May 2019

Cleveland Clinic
Familial Hypercholesterolemia: The Epitome of Personalized Medicine
Medical Grand Rounds
Cleveland, OH June 2019

XIV. Community Service

2006-2010	Arbor Clinic, free clinic operated by Stanford Medical Students: Attending Physician ~ one-half day per 1-2 mos. (volunteer)
2010-2017	Healthy Heart Week (volunteer): Presentation to local elementary school about the importance of a healthy lifestyle for heart health.
2011-2012	Mentor for at risk high school students, Woodside High School: Help to provide mentorship to two high school students who have medical career goals. At least one-half day per quarter.
2016-2017	Mentor to two high school students for Authentic Research Program organized by Palo Alto City School System
2021-2022	Corte Madera Middle School, volunteer girls basketball coach
2021-present	Board Member, Portola Valley AYSO (Youth Soccer)

XV. Trainees

Giuliano Lobos, Howard University Medical Student
5-7/2022
Giuliano Lobos worked in the lab as part of the Stanford CVI's Summer Research Program. With direct supervision of Dr. Ewa Bielczyk-Maczyńska – the staff scientist in the lab, his project focused on validating a screen for novel receptors involved in adipogenesis.

Dora Martelli, Senior High School Student
6-8/2022
Dora worked in the lab as a rising senior in high school and was part of the Stanford Institute of Medical Research (SIMR) Summer Program for High School Students. She worked under the guidance of Theresia Schnurr about exercising immortalized skeletal muscle cells in vitro using electrical pulse stimulation.

Theresia Schnurr, PhD, Postdoctoral researcher
11/20-
Theresia is a postdoctoral scholar from Denmark funded by a prestigious grant who is here for a planned three years working on the intersection of the genetics of insulin resistance and exercise/fitness. She is co-mentored with Dr. Euan Ashley

Peter Gustaffson, PhD, Postdoctoral researcher (formerly in Erik Ingelsson lab)
12/19-6/22
Peter is a postdoctoral scholar originally recruited from Sweden to Dr. Ingelsson's lab to study NAFLD. Funded by the Swedish Research Council grant. When Dr. Ingelsson took a leave of absence from Stanford, I became his primary mentor.

Ewa Bielczyk-Maczynska, PhD, Postdoctoral researcher (formerly in Erik Ingelsson lab)
12/19-

Ewa is studying the role of cell surface receptors in adipogenesis. She was recruited to Dr. Ingelsson's lab from the lab of Dr. Mary Teruel. When Dr. Ingelsson took a leave of absence from Stanford, I became her primary mentor. Ewa will stay on as a LSRP starting May 2022.

Jiehan Li, PhD, Postdoctoral researcher (formerly in Erik Ingelsson lab)
12/19-6/22
Jiehan is a talented postdoctoral fellow studying adipogenesis and insulin resistance, including using CRISPR screens. When Dr. Ingelsson took a leave of absence from Stanford, I became a co-mentor along with Dr. Thomas Quertermous.

Johanne Justesen, PhD, Postdoctoral researcher (formerly in Erik Ingelsson lab but co-Mentored with Dr. Manuel Rivas)
12/19-6/222
Johanne is a postdoctoral scholar from Denmark with extensive dry bench experience who is gaining an appreciation of wet bench functional studies to complement her other work. Presently mostly studying NAFLD. Splits her time in my lab and with Dr. Manny Rivas on dry bench projects within the UK Biobank

Laeya Najmi, PhD, Postdoctoral researcher (formerly in Erik Ingelsson lab but co-Mentored with Dr. Manuel Rivas)
12/19-4/22
Laeya was studying the genetic basis of insulin resistance using CRISPR approaches. Dr. Najmi will become a scientist at Sangamo Therapeutics.

Panjamaporn "Pam" Sangwung, PhD, Postdoctoral researcher
9/17-4/22
Pam was a postdoctoral scholar that came from Case Western Reserve University where she completed her PhD. She is studying the molecular mechanisms of *Nat1* induced insulin resistance. Dr. Sangwuang is now an Advisor at Molecular Pharmacology, Eli Lily and Company.

Cameron Cornn, Student Intern
6/20-8/20
Cameron is a premed student from Colorado who did a summer research (virtual) experience in the lab (UIM student)

Jay Singh, BA, Research Assistant
2019
Jay was a pre-med scholar who did a gap research experience with the Knowles lab in 2019. He assisted with mouse colony management and genotyping, as well as other assays. He will be attending UC Davis for medical school in the fall of 2020.

Karina Samuel-Gama, Student Intern and Research Assistant
2016-2017
Karina worked as a Laboratory Research Assistant in the Knowles and Quertermous labs from 2016 to 2017. She performed protein analyses to elucidate the role of genes FAM13A and NAT1

in insulin resistance. She also assisted with mouse colony management, mammalian cell culture, and quantitative protein and RNA assays (UIM student).

Wilmene Hercule, Student Intern and Research Assistant
2015
Willie was a HumBio intern and research assistant in the Knowles lab in 2015. She is now a Research Assistant/Clinical Anatomy Scholar at the Division of Clinical Anatomy at Stanford University School of Medicine, Department of Surgery (UIM student)

Vander Harris, BA, Research Coordinator
7/17-6/18
Vander is a Stanford Undergraduate student who worked to help coordinate our trial of statins and diabetes while he applies to medical school.
Left to study for MCAT and take additional courses for medical school. (UIM student)

Chelsea Harris, BA, Research Coordinator
10/16-7/17
Chelsea is a Stanford Undergraduate student who is working to help coordinate our trial of statins and diabetes while she applies to medical school.
Left to matriculate at Duke Medical School (UIM student)

Mohsen Fathzadeh, PhD, Postdoctoral researcher
9/15-12/19
Mohsen came to us from a very nice PhD at Yale where he examined *DRK1B* (NEJM publication). He worked on the hepatic specific effects of *NAT2* deficiency as well as on another promising candidate IR locus, *FAM13A*.
Left to take another postdoctoral appointment at Stanford (dry bench position)

Ivan Carcamo-Orive, PhD, Postdoctoral researcher
8/12-2/18
Ivan worked on the GENESIPS project, a large-scale effort to use human induced pluripotent stem cells to create model systems for the study of insulin resistance. In our lab he pioneered multiple techniques related to iPSC biology including endothelial cell differentiation and characterization. Co-mentored with Thomas Quertermous
Now senior scientist in the Knowles lab

Hadi Harati, MD, Postdoctoral researcher
7/15-7/19
Hadi was an Endocrinology fellow who worked with Dr. Ingelsson and I on Mendelian Randomization approaches to study links between diabetes and atrial fibrillation.
At the conclusion of his fellowship he took a job at Kaiser as an Endocrinologist

Indumathi Chennamsetty, PhD, Postdoctoral researcher
9/12-7/17
Indu worked on the functional characterization of NAT2 as an insulin sensitivity locus mostly using in vitro assays. Co-mentored with Thomas Quertermous

Left to take a job at Bayer

Qi Huang, MD, Visiting scholar
2/13-1/14
Qi is a trained endocrinologist from China who had returned to Wuhan University to get a PhD. She earned a prestigious scholarship to come to Stanford for a year to study insulin resistance. She worked in tandem with other lab members on projects related to the pathways underlying the effect of *NAT2* on insulin resistance.
Returned to China as MD

Mohammad Shahbazi, PhD, Postdoctoral researcher
9/13-5/16
Mohammad came to Stanford with a PhD in stem cell biology from Stanford. His project focused on the differentiation and characterization of iPSCs to adipocytes as well as understanding the role of insulin on iPSC growth, development and differentiation
Left to take another Postdoc at Stanford in Cardiothoracic surgery

Emily Youngblom, BA, Public Health Genomics Practicum
6/13-8/13
Emily came to Stanford to complete her "Practicum" as part of her curriculum to obtain a Master's degree in Public Health Genomics at the University of Washington. She worked on several projects related to Familial Hypercholesterolemia (FH) including drafting a "GeneReview" and a survey of schools of public health to determine if FH is a part of their curriculum.
Returned to genetic counseling school

Cynthia Li, High School Student, Summer Internship
June 2012
Cynthia was a rising high school junior that worked getting a "first lab experience". She learned from other members of the lab how to do basic techniques like RNA extraction, protein lysis, cell culture and Western blots.