

Biographical and Bibliographic Information

JOSHUA W. KNOWLES

A. Academic history:

Colleges and universities attended, degrees received, dates.

8/91-5/95	BA	History, Biology (<i>w/honors</i>) 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

Other study and research opportunities

Research Grants (*current*):

7/1/10-6/30/15	AHA National Fellow to Faculty Award, 10FTF3360005	SPO # 48273
	Identification and characterization of genetic determinants of insulin resistance	
	PI	
4/1/11-3/30/15	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
12/20/12-1/7/14	Stanford CVI Seed Grant	
	Hybrid genotyping of a well-phenotyped healthy control population as a community resource for exome studies	
	Co-PI w/ Jason Merker, MD, PhD (Genetics)	

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)

Research Grants (submitted or planned submission)

4/1/14-3/30/18 NIH/NHGRI RFA-HG-12-006
 Implementing cardiovascular clinical care algorithms including family history and genomics
 Co-Investigator co-PIs: Euan Ashley (Stanford), Dan Rader (Penn)

12/1/13-6/30/15 PCORI Patient-Powered Research Networks (PPRN)
 CAscade SCreening for Awareness and DETection of FH (CASCADE-FH) network
 Co-Investigator PI: Eric Peterson (Duke)

Research Grants (past)

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

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

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

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

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
4/1/08-4//109	Cardiovascular Institute Seed Grant, 2008 Genome-wide association study for determinants of insulin sensitivity Co-Investigator	PI: Thomas Quertermous, MD
6/5/08-6/4/09	Rosetta Inpharmatics LLC Genome-wide association study for determinants of insulin sensitivity Co-Investigator	PI: Thomas Quertermous, MD
5/1/10-4/30/11	Stanford CTSA Seed Grant, Innovation Award in Population Science Supported by NIH/NCRR CTSA <i>A pilot randomized trial of personalized genetics in preventive cardiology</i> PI	UL1 RR025744
10/1/11-9/30/12	Stanford CVI Seed Grant <i>A pilot randomized trial of personalized genetics in preventive cardiology</i> PI	1091650-310-GHAKD

Clinical Trials

STUDY: 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 70 of a planned 100 patients at Stanford
 Ongoing, recruiting patients.
 NCT ID: **NCT 00248079**

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
11/2012	Diplomate, American Board of Clinical Lipidology

Licensure

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

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

Academic positions:

8/16/10-6/30/11	Clinical Instructor, Division of Cardiovascular Medicine Stanford University School of Medicine, Stanford, CA
7/1/11-6/30/16	Instructor, Division of Cardiovascular Medicine

Stanford University School of Medicine, Stanford, CA

Non-Academic positions:

8/16/10-pres. Attending Physician, Stanford Center for Inherited Cardiovascular Disease
Stanford Hospital and Clinics, Stanford, CA

C. Public and professional service.

International committees and task forces:

National committees and task forces:

2012-pres. FH Foundation (Patient-led, advocacy group/charitable organization)
Chief Medical Officer
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; Participating in the AHA Scientific Statement on FH; Testifying at the FDA in favor of new therapies for homozygous FH (November, 2012); 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.

2013 Organizing Committee and co-Host
The Familial Hypercholesterolemia Summit: Awareness to Action
Annapolis, MD September 18-19, 2013

2013 Writing Group, American Heart Association
AHA Scientific Statement on Familial Hypercholesterolemia
Chair of writing group: Sam Gidding

Local committees and task forces:

2008-2009 Stanford Initiative in Human Genetics and Genomics
Organizer, campus-wide seminar series on human genetics topics

2011-pres. Pacific Lipid Association (part of the National Lipid Association) Familial Hypercholesterolemia (FH) working group

Community Service:

- 11/06-present Arbor Clinic, free clinic operated by Stanford Medical Students:
Attending Physician ~ one-half day per 1-2 mos. (volunteer)
- 2010-2013 Healthy Heart Week (volunteer): Presentation to local elementary school
about the importance of a healthy lifestyle for heart health.
- 10/11-10/12 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.

Editorial Experience:

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*

Assisted with reviews for:

- 2009-pres. *Circulation*
- 2009-pres. *New England Journal of Medicine*
- 2011-pres. *Journal of Clinical Investigation*

D. Post-degree honors and awards if any. Include major invited papers and addresses,
memberships in professional associations and learned societies, etc.

- 2003-2005 Stanford Clinical Investigator Pathway Program
*Program designed to mentor candidates with outstanding credentials for research
careers.*
- 2006-2007 The Future Leaders in CV Medical Research Program Fellowship Award
- 6/2007 Edwin Alderman Award for Excellence in Clinical Research
Awarded to the top research project by a Cardiology Fellow (judged by faculty)
- 6/2008 Edwin Alderman Award for Excellence in Clinical Research
Awarded to the top research project by a Cardiology Fellow (judged by faculty)
- 9/2012 Fellow, American College of Cardiology
- 11/2012 Fellow, American Heart Association

E. Publications

E.1. Peer-reviewed articles

E.1.A *Original research* contributions (H-factor 24 based on Google scholar 6/2013)

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 hemostatic 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 atherosclerosis: 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 fatty 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, Tuolimehto J, Quertermous 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 (in preparation w/ planned submission)*

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. 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 (*in revision*). [Helped to conceptualize idea for manuscript, contributed to editing manuscript].
40. 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].
41. 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.
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; the GENESIS consortium; the RISC consortium, Lehtimäki T, Liu CT, Loos RJ, McCarthy MI, Morris AD, Vasan RS, Spector TD, Teslovich TM, Tuomilehto J, Willems van Dijk K, 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 Randomisation Studies Do Not Support a Causal Role for Reduced Circulating Adiponectin Levels in Insulin Resistance and Type 2 Diabetes. *Diabetes*. 2013 Jul 8. [Epub ahead of print]. [Contributed reagents/genotyping data, assistance writing manuscript]

* co-first authors

E.1.B *Reviews and case reports*

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.

E.2. Non-peer-reviewed articles

1. **Knowles JW**, Maeda N. Dyslipidemia and hypertension: twin killers in renal vascular disease. *Am J Kidney Dis* 2001;37(6):1322-1323.

E.3. Books [0]

E.4. Book chapters [1]

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

E.5. Book reviews [0]

E.6. Abstracts

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
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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

E.7. Invited presentations

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, UY 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 Nov., 2007

4. *Genome Wide Association Studies of Cardiovascular Disease*
Stanford Biostatistics Seminar
Stanford, CA Jan., 2008
5. *The Athlete's Heart*
Senior Games of California
Stanford, CA Apr., 2010
6. *Genetics of Cardiovascular Disease*
Stanford School of Medicine, Introductory Epidemiology Course [Instructor: Rita Popat]
Stanford, CA Mar., 2010
7. Panelist, Speed Science
Stanford Cardiovascular Institute Member Retreat
Stanford, CA Sep., 2010
8. *Genome wide association study for determinants of insulin sensitivity*
Vanderbilt University, Cardiology Special Seminar
Nashville, TN Dec., 2010
9. *Genome wide association study for determinants of insulin sensitivity*
University of Pennsylvania, Cardiology Special Seminar
Philadelphia, PA Dec., 2010
10. *Personalized medicine and genomics*
Stanford School of Medicine, Introductory Epidemiology Course (*GENE210*)
Stanford, CA April, 2011
11. *A pilot randomized trial of personal genomics for preventive cardiology*
Stanford Medical School lecture (*MED223*)
Stanford, CA June, 2011
12. *Genetics of Cardiovascular Disease*
Stanford SIMR Summer Research Program
Stanford, CA June, 2011
13. *Atherogenesis*
Stanford EXPLORE Summer Research Program
Stanford, CA June, 2011
14. *Health and Wellness for the Busy Executive*
Stanford Executive Program, Stanford Graduate School of Business
Stanford, CA June, 2011
15. *The Heart*
Stanford School of Medicine, Lecture for 1st year medical students
Stanford, CA July, 2011
16. *Atherogenesis*

Stanford Cardiology Fellow Educational Lecture Series
Stanford, CA Fall, 2011

17. *Genetics of Cardiovascular Disease*
CME talk for Stanford Internal Medicine group
Stanford, CA July, 2011
18. *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
19. *Impacts of Genomics on Disease Treatment*
SAMMS Panel Discussion (School of Medicine initiative to foster cross disciplinary collaboration)
Stanford, CA Dec. 2011
20. *Randomized trial of personal genomics in preventive cardiology*
Stanford Cardiology Grand Rounds
Stanford, CA Jan., 2012
21. *Genetics of Cardiovascular Disease*
Stanford School of Medicine, Introductory Epidemiology Course [Instructor: Rita Popat]
Stanford, CA Feb., 2012
22. *Cardiovascular Genetics*
Apple Computers, Health Series
Cupertino, CA Mar., 2012
23. *A randomized trial of personal genomics for preventive cardiology: design and challenges*
American Heart Association National Meeting
Los Angeles, CA Nov., 2012
24. *Genetics of Cardiovascular Disease*
Stanford SIMR Summer Research Program
Stanford, CA June, 2012
25. *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
26. *Focus on FH*
Stanford Cardiology Grand Rounds
Stanford, CA Sep., 2012
27. *Focus on FH*
Kaiser Permanente San Francisco
San Francisco, CA Oct., 2012

28. *Focus on FH*
Stanford Internal Medicine Noon Conference
Stanford, CA Oct., 2012
29. *Approach to the Statin Intolerant Patient*
Stanford CME program
Stanford, CA Oct., 2012
30. *Focus on FH*
Medical Genetics Grand Rounds, Stanford
Stanford, CA Nov., 2012
31. *Cardiovascular Genetics*
Stanford School of Medicine, Introductory Epidemiology Course [Instructor: Rita Popat]
Stanford, CA Feb., 2013
32. *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 Feb., 2013
33. *Approach to the Statin Intolerant Patient*
Stanford Cardiology Grand Rounds, Stanford
Stanford, CA March, 2013
34. *Pre-hypertension and hypertension in the young adult*
Stanford Vaden Health, CME, Stanford
Stanford, CA April, 2013
35. *FH: Genetics, screening and treatment*
Preventive Cardiology Nurses Association National Meeting
Las Vegas, NV May, 2013
36. *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
37. *Approach to the Statin Intolerant Patient*
Stanford Internal Medicine Resident lecture series, Stanford and Palo Alto VA
Stanford, CA August, 2013
38. *What is it like to be a cardiologist?*
Stanford Explore Series (High school students)
Stanford, CA August, 2013
39. *Nuts and Bolts of Genetic Testing for FH*
National Lipid Association Regional Meeting, September, 2013
Baltimore, MD September, 2013

E.8. Patents N/A

F. Trainees

Ivan Carcamo-Orive, PhD, Postdoctoral researcher
8/12-

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

Indumathi Chennamsetty, PhD, Postdoctoral researcher
9/12-

Indu worked on the functional characterization of NAT2 as an insulin sensitivity locus mostly using in vitro assays. Co-mentored with Thomas Quertermous

Qi Huang, MD, Visiting scholar
2/13-2/14

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

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.

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.