

# Stanford

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## Joshua W. Knowles

Associate Professor of Medicine (Cardiovascular Medicine)  
Medicine - Cardiovascular Medicine

NIH Biosketch available Online

Curriculum Vitae available Online

### CLINICAL OFFICE (PRIMARY)

- **Cardiovascular Medicine**

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Falk Bldg CVRC MC 5406  
Stanford, CA 94305

**Tel** (650) 736-7878      **Fax** (650) 498-7452

### ACADEMIC CONTACT INFORMATION

- **Alternate Contact**

Tina Shen

**Email** [yshen9@stanford.edu](mailto:yshen9@stanford.edu)

## Bio

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### BIO

I am a physician-scientist and the overall theme my research has been the genetic basis of cardiovascular disease across the continuum from Discovery to the development of Model Systems to the Translation of these findings to the clinic and most recently to the Public Health aspect of genetics. I completed his MD-PhD at UNC with Prof. Nobuyo Maeda and Nobel Laureate Oliver Smithies. I did Internal Medicine residency and Cardiology fellowship training at Stanford working with Dr. Tom Quertermous. Currently my Discovery and basic translational efforts center on understanding the genetic basis of insulin resistance using GWAS studies coupled with exploration in model systems including iPSCs and CRISPR screens. My clinical translational focus is on Familial Hypercholesterolemia (FH) and I am the volunteer Chief Medical Advisor of the FH Foundation (FHF) which is a patient-led organization dedicated to increasing awareness of FH, identifying and treating patients with FH and screening family members to prevent deleterious outcomes. I helped lead the FHF efforts to establish a national patient registry (CASCADE FH), apply for an ICD10 code for FH and am now using cutting-edge “big-data” approaches to identify previously undiagnosed FH patients in electronic medical records (FIND FH). I have published over 90 papers with research projects currently funded by the National Institutes of Health, the American Diabetes Association, the American Heart Association and the Doris Duke Charitable Foundation.

### CLINICAL FOCUS

- Cardiology
- Insulin Resistance
- Familial Hypercholesterolemia
- Cardiovascular Disease
- Genetics
- Preventive cardiology
- Cholesterol
- Hypertriglyceridemia
- Polygenic risk
- statin intolerance

## **ACADEMIC APPOINTMENTS**

- Associate Professor - University Medical Line, Medicine - Cardiovascular Medicine
- Member, Bio-X
- Member, Cardiovascular Institute
- Member, Wu Tsai Human Performance Alliance
- Member, Maternal & Child Health Research Institute (MCHRI)

## **ADMINISTRATIVE APPOINTMENTS**

- Co-Section Chief, Metabolism and Insulin Signaling, Stanford Diabetes Research Center, (2017- present)
- Program Director, Cardiovascular Medicine Fellowship Program, (2018- present)
- Co-Director, Translational Investigator Program, (2017- present)
- Chief Research Advisor, The Family Heart Foundation, (2012- present)

## **HONORS AND AWARDS**

- National Innovative Research Award, AHA
- Dean's Fellowship, Stanford (2005)
- Clinical Scholar, Doris Duke Charitable Foundation (6/1/16-6/1/19)
- Fellowship Award, Future Leaders in CV Medicine (2006)
- Alderman Award for Excellence in Clinical Research, Stanford Cardiovascular Medicine (2008, 2009)
- Diplomate, National Lipid Association (2012)
- Fellow, AHA (2012)
- Fellow, ACC (2012)
- National Fellow to Faculty Transition Award, AHA (7/1/10-7/1/15)

## **BOARDS, ADVISORY COMMITTEES, PROFESSIONAL ORGANIZATIONS**

- Board Member, The FH Foundation (2012 - present)

## **PROFESSIONAL EDUCATION**

- Fellowship: Stanford University Cardiovascular Medicine Fellowship (2010) CA
- Residency: Stanford University Internal Medicine Residency (2005) CA
- Board Certification: Cardiovascular Disease, American Board of Internal Medicine (2010)
- Medical Education: University of North Carolina - Chapel Hill (2003) NC
- PhD, University of North Carolina, Chapel Hill , Genetics and Molecular Biology (2001)

## **COMMUNITY AND INTERNATIONAL WORK**

- The FH Foundation
- Arbor Free Clinic

## **LINKS**

- Stanford Center for Inherited Cardiovascular Disease: <http://familyheart.stanford.edu>
- The FH Foundation: <http://www.thefhfoundation.org>
- Stanford TIP: <https://medicine.stanford.edu/residency/tip.html>
- Stanford Cardiology Fellowship: <http://med.stanford.edu/cvmedicine/education/general-cardiology-fellowship.html>

- Get a Second Opinion: <https://stanfordhealthcare.org/second-opinion/overview.html>

## Research & Scholarship

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### CURRENT RESEARCH AND SCHOLARLY INTERESTS

The overall theme of my work is to understand the genetic basis of complex cardiovascular diseases such as coronary disease and insulin resistance. Currently, I am involved in genome wide association (GWA) studies of coronary disease through the NIH-funded ADVANCE study and of insulin resistance through the international GENESIS project. After using the GWA approach to discover and validate interesting candidate genes, I hope to explore the biology underlying these genes and pathways using cell culture and *in vivo* model systems. I am also using iPSC technology (induced pluripotent stem cells) to develop model systems for the study of insulin resistance through the NIH-funded GENESiPS project.

I also have a strong interest in developing tools to help translate newly emerging genetic data into clinical practice and am currently the PI of a randomized trial where we are examining whether we can improve patient coronary disease risk factor profiles by giving them information about their inherited risk of coronary disease.

Clinically, I am interested in the care of patients with inherited cardiovascular conditions. In particular I am focused on individuals with Familial Hypercholesterolemia. I also have an interest in treating Hypertrophic Cardiomyopathy. I take care of these patients in the Stanford Center for Inherited Cardiovascular Disease.

### CLINICAL TRIALS

- Personal Genomics for Preventive Cardiology, Not Recruiting
- Relationship Between Insulin Resistance and Statin Induced Type 2 Diabetes, and Integrative Personal Omics Profiling, Not Recruiting

## Publications

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### PUBLICATIONS

- **Development and utility of a clinical research informatics application for participant recruitment and workflow management for a return of results pilot trial in familial hypercholesterolemia in the Million Veteran Program.** *JAMIA open*  
Brunette, C. A., Yi, T., Danowski, M. E., Cardellino, M., Harrison, A., Assimes, T. L., Knowles, J. W., Christensen, K. D., Sturm, A. C., Sun, Y. V., Hui, Q., Pyarajan, S., Shi, et al  
2024; 7 (1): ooae020
- **Lac-Phe mediates the effects of metformin on food intake and body weight.** *Nature metabolism*  
Xiao, S., Li, V. L., Lyu, X., Chen, X., Wei, W., Abbasi, F., Knowles, J. W., Tung, A. S., Deng, S., Tiwari, G., Shi, X., Zheng, S., Farrell, et al  
2024
- **Generation of two induced pluripotent stem cell lines from healthy patients of African American ancestry.** *Stem cell research*  
Mullen, M., Kojic, A., Alamana, C., Canel, G., Lai, C., Knowles, J. W., Wu, J. C.  
2024; 76: 103322
- **A functional genomic framework to elucidate novel causal non-alcoholic fatty liver disease genes.** *medRxiv : the preprint server for health sciences*  
Saliba-Gustafsson, P., Justesen, J. M., Ranta, A., Sharma, D., Bielczyk-Maczynska, E., Li, J., Najmi, L. A., Apodaka, M., Aspichueta, P., Björck, H. M., Eriksson, P., Franco-Cereceda, A., Gloudemanns, et al  
2024
- **Trends of Lipid Concentrations, Awareness, Evaluation, and Treatment in Severe Dyslipidemia in US Adults.** *Mayo Clinic proceedings*  
Shetty, N. S., Gaonkar, M., Patel, N., Knowles, J. W., Natarajan, P., Arora, G., Arora, P.  
2024
- **Impact of Virtual Interviewing on Cardiovascular Fellowship Applicant Diversity: Insights From 2 Academic Programs.** *Journal of the American Heart Association*  
Witting, C., Knowles, J. W., DeFaria Yeh, D., Beyene, T. J., Gummipundi, S. E., Heidenreich, P. A., Yong, C. M.  
2023: e030255

- **RNA Interference Therapy Targeting Apolipoprotein C-III in Hypertriglyceridemia. NEJM evidence**  
Gaudet, D., Clifton, P., Sullivan, D., Baker, J., Schwabe, C., Thackwray, S., Scott, R., Hamilton, J., Given, B., Melquist, S., Zhou, R., Chang, T., San Martin, et al  
2023; 2 (12): EVIDoa2200325
- **USPSTF Recommendation on Screening for Lipid Disorders in Children and Adolescents. JAMA**  
Wilemon, K., Knowles, J. W., Baum, S. J.  
2023; 330 (20): 2024
- **Lac-Phe mediates the anti-obesity effect of metformin. bioRxiv : the preprint server for biology**  
Xiao, S., Li, V. L., Lyu, X., Chen, X., Wei, W., Abbasi, F., Knowles, J. W., Deng, S., Tiwari, G., Shi, X., Zheng, S., Farrell, L., Chen, et al  
2023
- **Principles for Health Information Collection, Sharing, and Use: A Policy Statement From the American Heart Association CIRCULATION**  
Spector-Bagdady, K., Armondas, A. A., Arnaout, R., Hall, J. L., McSwain, B., Knowles, J. W., Price, W., Rawat, D. B., Riegel, B., Wang, T. Y., Wiley, K., Chung, M. K., Amer Heart Assoc Advocacy Coor  
2023; 148 (13): 1061-1069
- **RNA interference targeting ANGPTL3 for triglyceride and cholesterol lowering: phase 1 basket trial cohorts. Nature medicine**  
Watts, G. F., Schwabe, C., Scott, R., Gladding, P. A., Sullivan, D., Baker, J., Clifton, P., Hamilton, J., Given, B., Melquist, S., Zhou, R., Chang, T., San Martin, et al  
2023
- **ARO-ANG3, AN INVESTIGATIONAL RNAI THERAPEUTIC, DECREASES SERUM LDL-CHOLESTEROL, APOLIPOPROTEIN B, AND ANGIOPOIETIN-LIKE PROTEIN 3 IN PATIENTS WITH HOMOZYGOUS FAMILIAL HYPERCHOLESTEROLAEMIA**  
Raal, F., Bergeron, J., Watts, G., Gaudet, D., Sullivan, D., Turner, T., Hegele, R., Ballantyne, C., Knowles, J., Goldberg, I., Fu, R., Alagarsamy, S., Hamilton, et al  
ELSEVIER IRELAND LTD.2023
- **A single-cell CRISPRi platform for characterizing candidate genes relevant to metabolic disorders in human adipocytes. American journal of physiology. Cell physiology**  
Bielczyk-Maczynska, E., Sharma, D., Blencowe, M., Saliba Gustafsson, P., Gloudemans, M. J., Yang, X., Carcamo-Orive, I., Wabitsch, M., Svensson, K. J., Park, C. Y., Quertermous, T., Knowles, J. W., Li, et al  
2023
- **Plasma proteomic signatures of a direct measure of insulin sensitivity in two population cohorts. Diabetologia**  
Zanetti, D., Stell, L., Gustafsson, S., Abbasi, F., Tsao, P. S., Knowles, J. W., Zethelius, B., Ärnlöv, J., Balkau, B., Walker, M., Lazzeroni, L. C., Lind, L., Petrie, et al  
2023
- **Single-cell transcriptome dataset of human and mouse in vitro adipogenesis models. Scientific data**  
Li, J., Jin, C., Gustafsson, S., Rao, A., Wabitsch, M., Park, C. Y., Quertermous, T., Knowles, J. W., Bielczyk-Maczynska, E.  
2023; 10 (1): 387
- **Contemporary Homozygous Familial Hypercholesterolemia in the United States: Insights From the CASCADE FH Registry. Journal of the American Heart Association**  
Cuchel, M., Lee, P. C., Hudgins, L. C., Duell, P. B., Ahmad, Z., Baum, S. J., Linton, M. F., de Ferranti, S. D., Ballantyne, C. M., Larry, J. A., Hemphill, L. C., Kindt, I., Gidding, et al  
2023: e029175
- **Single-cell transcriptome dataset of human and mouse in vitro adipogenesis models. bioRxiv : the preprint server for biology**  
Li, J., Jin, C., Gustafsson, S., Rao, A., Wabitsch, M., Park, C. Y., Quertermous, T., Bielczyk-Maczynska, E., Knowles, J. W.  
2023
- **The relationship between insulin resistance and ion mobility lipoprotein fractions. American journal of preventive cardiology**  
Rowland, C. M., Abbasi, F., Schiffman, D., Knowles, J. W., McPhaul, M. J.  
2023; 13: 100457
- **G protein-coupled receptor 151 regulates glucose metabolism and hepatic gluconeogenesis. Nature communications**  
Bielczyk-Maczynska, E., Zhao, M., Zushin, P. H., Schnurr, T. M., Kim, H. J., Li, J., Nallagatla, P., Sangwung, P., Park, C. Y., Conn, C., Stahl, A., Svensson, K. J., Knowles, et al  
2022; 13 (1): 7408

- **Analysis of Transcriptional Variability in a Large Human iPSC Library Reveals Genetic and Non-genetic Determinants of Heterogeneity (vol 20, pg 518, 2017) *CELL STEM CELL***  
Carcamo-Orive, I., Hoffman, G. E., Cundiff, P., Beckmann, N. D., D'Souza, S. L., Knowles, J. W., Patel, A., Hendry, C., Papatsenko, D., Abbasi, F., Reaven, G. M., Whalen, S., Lee, et al  
2022; 29 (10): 1505
- **Chronic liver disease-related mortality in diabetes before and during the COVID-19 in the United States. *Digestive and liver disease : official journal of the Italian Society of Gastroenterology and the Italian Association for the Study of the Liver***  
Kim, D., Alshuwaykh, O., Dennis, B. B., Cholankeril, G., Knowles, J. W., Ahmed, A.  
2022
- **Analysis of Insulin Resistance Among Children and Adolescents in Slovenia With Hypercholesterolemia After Treatment With Statins. *JAMA network open***  
Groselj, U., Sikonja, J., Mlinaric, M., Kotnik, P., Battelino, T., Knowles, J. W.  
2022; 5 (9): e2231097
- **STATIN THERAPY IS NOT ASSOCIATED WITH INSULIN RESISTANCE IN CHILDREN AND ADOLESCENTS WITH FAMILIAL HYPERCHOLESTEROLAEMIA**  
Groselj, U., Sikonja, J., Mlinaric, M., Kotnik, P., Battelino, T., Knowles, J. W.  
ELSEVIER IRELAND LTD.2022: E173
- **Creation of the American Heart Association Journals' Equity, Diversity, and Inclusion Editorial Board: The Next Step to Achieving the 2024 Impact Goal. *Circulation***  
Lewis, E. F., Beaty, C., Boltze, J., Breathett, K., Clair, W. K., de Las Fuentes, L., Essien, U. R., Goodell, H., Hinson, H. E., Kershaw, K. N., Knowles, J. W., Mazimba, S., Mujahid, et al  
2022: 101161CIRCULATIONAHA122061450
- **Apolipoprotein B, Residual Cardiovascular Risk After Acute Coronary Syndrome, and Effects of Alirocumab. *Circulation***  
Hagström, E., Steg, P. G., Szarek, M., Bhatt, D. L., Bittner, V. A., Danchin, N., Diaz, R., Goodman, S. G., Harrington, R. A., Jukema, J. W., Liberopoulos, E., Marx, N., McGinniss, et al  
2022: 101161CIRCULATIONAHA121057807
- **Generation of two iPSC lines from hypertrophic cardiomyopathy patients carrying MYBPC3 and PRKAG2 variants. *Stem cell research***  
Manhas, A., Jahng, J. W., Vera, C. D., Shenoy, S. P., Knowles, J. W., Wu, J. C.  
2022; 61: 102774
- **Integration of genetic colocalizations with physiological and pharmacological perturbations identifies cardiometabolic disease genes. *Genome medicine***  
Gloudemans, M. J., Balliu, B., Nachun, D., Schnurr, T. M., Durrant, M. G., Ingelsson, E., Wabitsch, M., Quertermous, T., Montgomery, S. B., Knowles, J. W., Carcamo-Orive, I.  
2022; 14 (1): 31
- **Integration of genetic colocalizations with physiological and pharmacological perturbations identifies cardiometabolic disease genes**  
Gloudemans, M. J., Balliu, B., Nachun, D., Durrant, M. G., Ingelsson, E., Wabitsch, M., Quertermous, T., Montgomery, S. B., Knowles, J., Carcamo-Orive, I.  
W B SAUNDERS CO-ELSEVIER INC.2022: S24-S25
- **A guide for the diagnosis of rare and undiagnosed disease: beyond the exome. *Genome medicine***  
Marwaha, S., Knowles, J. W., Ashley, E. A.  
2022; 14 (1): 23
- **Online Patient Education Materials Related to Lipoprotein(a): Readability Assessment. *Journal of medical Internet research***  
Pearson, K., Ngo, S., Ekpo, E., Sarraju, A., Baird, G., Knowles, J., Rodriguez, F.  
1800; 24 (1): e31284
- **G protein-coupled receptor 151 regulates glucose metabolism and hepatic gluconeogenesis *Nature Communications***  
Bielczyk-Maczynska, E., Zhao, M., Zushin, P. H., Schnurr, T. M., Kim, H., Li, J., Sangwung, P., Nallagatla, P., Park, C., Cornn, C., Stahl, A., Svensson, K. J., Knowles, et al  
2022
- **Interactions of physical activity, muscular fitness, adiposity, and genetic risk for NAFLD. *Hepatology communications***  
Schnurr, T. M., Katz, S. F., Justesen, J. M., O'Sullivan, J. W., Saliba-Gustafsson, P., Assimes, T. L., Carcamo-Orive, I., Ahmed, A., Ashley, E. A., Hansen, T., Knowles, J. W.

2022

- **Ultrarapid Nanopore Genome Sequencing in a Critical Care Setting.** *The New England journal of medicine*  
Gorzynski, J. E., Goenka, S. D., Shafin, K., Jensen, T. D., Fisk, D. G., Grove, M. E., Spiteri, E., Pesout, T., Monlong, J., Baid, G., Bernstein, J. A., Ceresnak, S., Chang, et al  
2022
- **Coronary Artery Disease Risk of Familial Hypercholesterolemia Genetic Variants Independent of Clinically Observed Longitudinal Cholesterol Exposure.** *Circulation. Genomic and precision medicine*  
Clarke, S. L., Tcheandjieu, C., Hilliard, A. T., Lee, M., Lynch, J., Chang, K. M., Miller, D., Knowles, J. W., O'Donnell, C., Tsao, P., Rader, D. J., Wilson, P. W., Sun, et al  
2022: CIRCGEN121003501
- **The Clinical Genome Resource (ClinGen) Familial Hypercholesterolemia Variant Curation Expert Panel consensus guidelines for LDLR variant classification.** *Genetics in medicine : official journal of the American College of Medical Genetics*  
Chora, J. R., Iacocca, M. A., Tichy, L., Wand, H., Kurtz, C. L., Zimmermann, H., Leon, A., Williams, M., Humphries, S. E., Hooper, A. J., Trinder, M., Brunham, L. R., Costa Pereira, et al  
1800
- **Signaling defects associated with insulin resistance in non-diabetic and diabetic individuals and modification by sex.** *The Journal of clinical investigation*  
Haider, N., Lebastchi, J., Jayavelu, A. K., Batista, T. M., Pan, H., Dreyfuss, J. M., Carcamo-Orive, I., Knowles, J. W., Mann, M., Kahn, C. R.  
2021
- **Statins Are Associated With Increased Insulin Resistance and Secretion.** *Arteriosclerosis, thrombosis, and vascular biology*  
Abbasi, F., Lamendola, C., Harris, C. S., Harris, V., Tsai, M., Tripathi, P., Abbas, F., Reaven, G., Reaven, P., Snyder, M. P., Kim, S. H., Knowles, J. W.  
2021: ATVBAHA121316159
- **Genetics of Type 2 Diabetes: Opportunities for Precision Medicine: JACC Focus Seminar.** *Journal of the American College of Cardiology*  
Kim, D. S., Gloyn, A. L., Knowles, J. W.  
2021; 78 (5): 496-512
- **Lipoprotein(a) and Benefit of PCSK9 Inhibition in Patients With Nominally Controlled LDL Cholesterol** *JOURNAL OF THE AMERICAN COLLEGE OF CARDIOLOGY*  
Schwartz, G. G., Szarek, M., Bittner, V. A., Diaz, R., Goodman, S. G., Jukema, J., Landmesser, U., Lopez-Jaramillo, P., Manvelian, G., Pordy, R., Scemama, M., Sinnaeve, P. R., White, et al  
2021; 78 (5): 421-433
- **Diverse Racial/Ethnic Group Underreporting and Underrepresentation in High-Impact Cholesterol Treatment Trials.** *Circulation*  
Saraju, A., Valencia, A., Knowles, J. W., Maron, D. J., Rodriguez, F.  
2021; 143 (24): 2409-2411
- **Health disparities in cardiometabolic risk among Black and Hispanic youth in the United States.** *American journal of preventive cardiology*  
Katz, S. F., Rodriguez, F., Knowles, J. W.  
2021; 6: 100175
- **Relation of Lipoprotein(a) Levels to Incident Type 2 Diabetes and Modification by Alirocumab Treatment** *DIABETES CARE*  
Schwartz, G. G., Szarek, M., Bittner, V. A., Bhatt, D. L., Diaz, R., Goodman, S. G., Jukema, J., Loy, M., Manvelian, G., Pordy, R., White, H. D., Steg, P., ODYSSEY OUTCOMES Comm Investigator  
2021; 44 (5): 1219-1227
- **Generation of two heterozygous MYBPC3 mutation-carrying human iPSC lines, SCVIi001-A and SCVIi002-A, for modeling hypertrophic cardiomyopathy.** *Stem cell research*  
Liu, L., Shenoy, S. P., Jahng, J. W., Liu, Y., Knowles, J. W., Zhuge, Y., Wu, J. C.  
2021; 53: 102279
- **Identification of rare and common regulatory variants in pluripotent cells using population-scale transcriptomics.** *Nature genetics*  
Bonder, M. J., Smail, C., Gloudemans, M. J., Fresard, L., Jakubosky, D., D'Antonio, M., Li, X., Ferraro, N. M., Carcamo-Orive, I., Mirauta, B., Seaton, D. D., Cai, N., Vakili, et al  
2021
- **Validation of an Integrated Risk Tool, Including Polygenic Risk Score, for Atherosclerotic Cardiovascular Disease in Multiple Ethnicities and Ancestries.** *The American journal of cardiology*

Weale, M. E., Riveros-Mckay, F., Selzam, S., Seth, P., Moore, R., Tarran, W. A., Gradovich, E., Giner-Delgado, C., Palmer, D., Wells, D., Saffari, A., Sivley, R. M., Lachapelle, et al  
2021

**• RECOMMENDATIONS FOR LDLR VARIANT INTERPRETATION BY THE CLINGEN'S FAMILIAL HYPERCHOLESTEROLEMIA EXPERT PANEL**

Chora, J., Iacocca, M., Tichy, L., Wand, H., Kurtz, L. C., Zimmermann, H., Meredith, A., Williams, M., Humphries, S. E., Hooper, A. J., Brunham, L., Pereira, A., Chen, et al

LIPPINCOTT WILLIAMS & WILKINS.2021

**• The need for polygenic score reporting standards in evidence-based practice: lipid genetics use case. *Current opinion in lipidology***

Wand, H. n., Knowles, J. W., Clarke, S. L.  
2021

**• An integrated approach to identify environmental modulators of genetic risk factors for complex traits. *American journal of human genetics***

Balliu, B., Carcamo-Orive, I., Gloudemans, M. J., Nachun, D. C., Durrant, M. G., Gazal, S., Park, C. Y., Knowles, D. A., Wabitsch, M., Quertermous, T., Knowles, J. W., Montgomery, S. B.  
2021

**• Isthmin-1 is an adipokine that promotes glucose uptake and improves glucose tolerance and hepatic steatosis. *Cell metabolism***

Jiang, Z., Zhao, M., Voilquin, L., Jung, Y., Aikio, M. A., Sahai, T., Dou, F. Y., Roche, A. M., Carcamo-Orive, I., Knowles, J. W., Wabitsch, M., Appel, E. A., Maikawa, et al  
2021

**• Predictive network modeling in human induced pluripotent stem cells identifies key driver genes for insulin responsiveness. *PLoS computational biology***

Carcamo-Orive, I., Henrion, M. Y., Zhu, K., Beckmann, N. D., Cundiff, P., Moein, S., Zhang, Z., Alamprese, M., D'Souza, S. L., Wabitsch, M., Schadt, E. E., Quertermous, T., Knowles, et al  
2020; 16 (12): e1008491

**• SPECIFICATION OF ACMG/AMP GUIDELINES FOR STANDARDIZED VARIANT INTERPRETATION IN FAMILIAL HYPERCHOLESTEROLEMIA: ON BEHALF OF THE CLINGEN FH VARIANT CURATION EXPERT PANEL**

Iacocca, M. A., Chora, J. R., Freiberger, T., Carrie, A., Sijbrands, E. J., Wand, H., Williams, M., Zimmermann, H., Leon, A., Kurtz, C. L., Tichy, L., Alves, A. C., Wang, et al  
ELSEVIER IRELAND LTD.2020: E5

**• RNAi inhibition of angiopoietin-like protein 3 (ANGPTL3) with ARO-ANG3 mimics the lipid and lipoprotein profile of familial combined hypolipidemia**

Watts, G. F., Schwabe, C., Scott, R., Gladding, P., Sullivan, D., Baker, J., Clifton, P., Hamilton, J., Given, B., San Martin, J., Melquist, S., Knowles, J. W., Goldberg, et al  
OXFORD UNIV PRESS.2020: 3331

**• RNA interference targeting apolipoprotein C-III with ARO-APOC3 in healthy volunteers mimics lipid and lipoprotein findings seen in subjects with inherited apolipoprotein C-III deficiency**

Schwabe, C., Scott, R., Sullivan, D., Baker, J., Clifton, P., Hamilton, J., Given, B., San Martin, J., Melquist, S., Watts, G. F., Goldberg, Knowles, J. W., Hegele, R., et al  
OXFORD UNIV PRESS.2020: 3330

**• Children with Heterozygous Familial Hypercholesterolemia in the United States: Data from the CASCADE FH Registry. *The Journal of pediatrics***

de Ferranti, S. D., Shrader, P., Linton, M. F., Knowles, J. W., Hudgins, L. C., Benuck, I., Kindt, I., O'Brien, E. C., Peterson, A. L., Ahmad, Z. S., Clauss, S., Duell, P. B., Shapiro, et al  
2020

**• Expression of Glycolytic Enzymes in Induced Pluripotent Stem Cells, Derived Megakaryocytes, and Endothelial Cells**

Chandra, A., Vaidya, D., Chang, R., Knowles, J., Carcamo-Orive, I., Kammers, K., Martin, J., Taub, M., Hoyle, D., Gao, Y., Yanek, L., Kral, B., Faraday, et al  
LIPPINCOTT WILLIAMS & WILKINS.2020

**• Discovery and quality analysis of a comprehensive set of structural variants and short tandem repeats. *Nature communications***

Jakubosky, D., Smith, E. N., D'Antonio, M., Jan Bonder, M., Young Greenwald, W. W., D'Antonio-Chronowska, A., Matsui, H., i2QTL Consortium, Stegle, O., Montgomery, S. B., DeBoever, C., Frazer, K. A., Bonder, M. J., et al  
2020; 11 (1): 2928

**• Properties of structural variants and short tandem repeats associated with gene expression and complex traits. *Nature communications***

Jakubosky, D., D'Antonio, M., Bonder, M. J., Smail, C., Donovan, M. K., Young Greenwald, W. W., Matsui, H., i2QTL Consortium, D'Antonio-Chronowska, A., Stegle, O., Smith, E. N., Montgomery, S. B., DeBoever, C., et al  
2020; 11 (1): 2927

● **Insulin Resistance and Mitochondrial Dysfunction Mediated by Nat1 Deficiency**

Sangwung, P., Fathzadeh, M., Knowles, J.  
AMER DIABETES ASSOC.2020

● **Gaps in Dyslipidemia Care Among Working-Aged Individuals With Employer-Sponsored Health Care.** *Journal of the American Heart Association*

Shiffman, D., Louie, J. Z., Devlin, J. J., Knowles, J. W., McPhaul, M. J.  
2020: e015807

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