Joshua W Knowles

List of Publications by Citations

102 papers	8,179 citations	34 h-index	90 g-index
117 ext. papers	9,838 ext. citations	8.4 avg, IF	5.45 L-index

#	Paper	IF	Citations
102	Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. *Nature Genetics*, **2010**, 42, 937-48	36.3	2267
101	Large-scale association analysis identifies 13 new susceptibility loci for coronary artery disease. *Nature Genetics*, **2011**, 43, 333-8	36.3	1394
100	Clinical assessment incorporating a personal genome. *Lancet, The*, **2010**, 375, 1525-35	40	565
99	Human induced pluripotent stem cell-derived cardiomyocytes recapitulate the predilection of breast cancer patients to doxorubicin-induced cardiotoxicity. *Nature Medicine*, **2016**, 22, 547-56	50.5	425
98	The Agenda for Familial Hypercholesterolemia: A Scientific Statement From the American Heart Association. *Circulation*, **2015**, 132, 2167-92	16.7	377
97	Impact of type 2 diabetes susceptibility variants on quantitative glycemic traits reveals mechanistic heterogeneity. *Diabetes*, **2014**, 63, 2158-71	0.9	235
96	Clinical Genetic Testing for Familial Hypercholesterolemia: JACC Scientific Expert Panel. *Journal of the American College of Cardiology*, **2018**, 72, 662-680	15.1	215
95	Detailed physiologic characterization reveals diverse mechanisms for novel genetic Loci regulating glucose and insulin metabolism in humans. *Diabetes*, **2010**, 59, 1266-75	0.9	211
94	Analysis of Transcriptional Variability in a Large Human iPSC Library Reveals Genetic and Non-genetic Determinants of Heterogeneity. *Cell Stem Cell*, **2017**, 20, 518-532.e9	18	164
93	Genetic evidence for a normal-weight "metabolically obese" phenotype linking insulin resistance, hypertension, coronary artery disease, and type 2 diabetes. *Diabetes*, **2014**, 63, 4369-77	0.9	131
92	Treatment Gaps in Adults With Heterozygous Familial Hypercholesterolemia in the United States: Data From the CASCADE-FH Registry. *Circulation: Cardiovascular Genetics*, **2016**, 9, 240-9		126
91	Phased whole-genome genetic risk in a family quartet using a major allele reference sequence. *PLoS Genetics*, **2011**, 7, e1002280	6	112
90	Association of Statin Adherence With Mortality in Patients With Atherosclerotic Cardiovascular Disease. *JAMA Cardiology*, **2019**, 4, 206-213	16.2	108
89	Cascade Screening for Familial Hypercholesterolemia and the Use of Genetic Testing. *JAMA - Journal of the American Medical Association*, **2017**, 318, 381-382	27.4	100
88	Mendelian randomization studies do not support a causal role for reduced circulating adiponectin levels in insulin resistance and type 2 diabetes. *Diabetes*, **2013**, 62, 3589-98	0.9	95
87	Association Between Intensity of Statin Therapy and Mortality in Patients With Atherosclerotic Cardiovascular Disease. *JAMA Cardiology*, **2017**, 2, 47-54	16.2	92
86	Using Genetic Variants to Assess the Relationship Between Circulating Lipids and Type 2 Diabetes. *Diabetes*, **2015**, 64, 2676-84	0.9	83

85	Cardiovascular disease: The rise of the genetic risk score. *PLoS Medicine*, **2018**, 15, e1002546	11.6	81
84	Identification and validation of N-acetyltransferase 2 as an insulin sensitivity gene. *Journal of Clinical Investigation*, **2015**, 125, 1739-51	15.9	67
83	Rationale and design of the familial hypercholesterolemia foundation CAscade SCreening for Awareness and DEtection of Familial Hypercholesterolemia registry. *American Heart Journal*, **2014**, 167, 342-349.e17	4.9	63
82	ClinVar database of global familial hypercholesterolemia-associated DNA variants. *Human Mutation* , **2018**, 39, 1631-1640	4.7	55
81	Cardiopulmonary responses and prognosis in hypertrophic cardiomyopathy: a potential role for comprehensive noninvasive hemodynamic assessment. *JACC: Heart Failure*, **2015**, 3, 408-418	7.9	51
80	Genetic susceptibility to peripheral arterial disease: a dark corner in vascular biology. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2007**, 27, 2068-78	9.4	49
79	Association of polymorphisms in platelet and hemostasis system genes with acute myocardial infarction. *American Heart Journal*, **2007**, 154, 1052-8	4.9	43
78	Measurement of insulin-mediated glucose uptake: direct comparison of the modified insulin suppression test and the euglycemic, hyperinsulinemic clamp. *Metabolism: Clinical and Experimental* , **2013**, 62, 548-53	12.7	42
77	US physician practices for diagnosing familial hypercholesterolemia: data from the CASCADE-FH registry. *Journal of Clinical Lipidology*, **2016**, 10, 1223-9	4.9	41
76	Prevalence and clinical correlates of right ventricular dysfunction in patients with hypertrophic cardiomyopathy. *American Journal of Cardiology*, **2014**, 113, 361-7	3	41
75	Simple, standardized incorporation of genetic risk into non-genetic risk prediction tools for complex traits: coronary heart disease as an example. *Frontiers in Genetics*, **2014**, 5, 254	4.5	39
74	Health disparities among adult patients with a phenotypic diagnosis of familial hypercholesterolemia in the CASCADE-FH patient registry. *Atherosclerosis*, **2017**, 267, 19-26	3.1	36
73	Maternal Midpregnancy Glucose Levels and Risk of Congenital Heart Disease in Offspring. *JAMA Pediatrics*, **2015**, 169, 1112-6	8.3	36
72	The role of registries and genetic databases in familial hypercholesterolemia. *Current Opinion in Lipidology*, **2017**, 28, 152-160	4.4	35
71	Longitudinal low density lipoprotein cholesterol goal achievement and cardiovascular outcomes among adult patients with familial hypercholesterolemia: The CASCADE FH registry. *Atherosclerosis* , **2019**, 289, 85-93	3.1	35
70	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**, 182, 97-102	4.9	34
69	Finding missed cases of familial hypercholesterolemia in health systems using machine learning. *Npj Digital Medicine*, **2019**, 2, 23	15.7	30
68	Mitochondrial Dysfunction, Insulin Resistance, and Potential Genetic Implications. *Endocrinology*, **2020**, 161,	4.8	30

67	Impact of a Genetic Risk Score for Coronary Artery Disease on Reducing Cardiovascular Risk: A Pilot Randomized Controlled Study. *Frontiers in Cardiovascular Medicine*, **2017**, 4, 53	5.4	29
66	Access to Nonstatin Lipid-Lowering Therapies in Patients at High Risk of Atherosclerotic Cardiovascular Disease. *Circulation*, **2017**, 135, 2204-2206	16.7	28
65	Large-Scale Phenome-Wide Association Study of Variants Demonstrates Protection Against Ischemic Stroke. *Circulation Genomic and Precision Medicine*, **2018**, 11, e002162	5.2	27
64	Impact of race/ethnicity on insulin resistance and hypertriglyceridaemia. *Diabetes and Vascular Disease Research*, **2019**, 16, 153-159	3.3	24
63	Precision screening for familial hypercholesterolaemia: a machine learning study applied to electronic health encounter data. *The Lancet Digital Health*, **2019**, 1, e393-e402	14.4	24
62	Failure to replicate an association of SNPs in the oxidized LDL receptor gene (OLR1) with CAD. *BMC Medical Genetics*, **2008**, 9, 23	2.1	24
61	Nat1 Deficiency Is Associated with Mitochondrial Dysfunction and Exercise Intolerance in Mice. *Cell Reports*, **2016**, 17, 527-540	10.6	24
60	Body composition and atrial fibrillation: a Mendelian randomization study. *European Heart Journal*, **2019**, 40, 1277-1282	9.5	23
59	Randomized trial of personal genomics for preventive cardiology: design and challenges. *Circulation: Cardiovascular Genetics*, **2012**, 5, 368-76		23
58	Prevalence and prognostic role of right ventricular involvement in stress-induced cardiomyopathy. *Journal of Cardiac Failure*, **2015**, 21, 419-425	3.3	22
57	Polygenic risk scores in coronary artery disease. *Current Opinion in Cardiology*, **2019**, 34, 435-440	2.1	18
56	FAM13A affects body fat distribution and adipocyte function. *Nature Communications*, **2020**, 11, 1465	17.4	17
55	Impact of Septal Reduction on Left Atrial Size and Diastole in Hypertrophic Cardiomyopathy. *Echocardiography*, **2016**, 33, 686-94	1.5	17
54	Frequency of Statin Use in Patients With Low-Density Lipoprotein Cholesterol ≥190 mg/dl from the Veterans Affairs Health System. *American Journal of Cardiology*, **2018**, 122, 756-761	3	17
53	First Trimester Plasma Glucose Values in Women without Diabetes are Associated with Risk for Congenital Heart Disease in Offspring. *Journal of Pediatrics*, **2018**, 195, 275-278	3.6	16
52	Identification of rare and common regulatory variants in pluripotent cells using population-scale transcriptomics. *Nature Genetics*, **2021**, 53, 313-321	36.3	16
51	Genetic Testing and Risk Scores: Impact on Familial Hypercholesterolemia. *Frontiers in Cardiovascular Medicine*, **2019**, 6, 5	5.4	15
50	Induced Pluripotent Stem Cell-Derived Endothelial Cells in Insulin Resistance and Metabolic Syndrome. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2017**, 37, 2038-2042	9.4	14

49	Association of insulin resistance, from mid-life to late-life, with aortic stiffness in late-life: the Atherosclerosis Risk in Communities Study. *Cardiovascular Diabetology*, **2020**, 19, 11	8.7	12
48	Hypertriglyceridemia: A simple approach to identify insulin resistance and enhanced cardio-metabolic risk in patients with prediabetes. *Diabetes Research and Clinical Practice*, **2016**, 120, 156-61	7.4	12
47	Discovery and quality analysis of a comprehensive set of structural variants and short tandem repeats. *Nature Communications*, **2020**, 11, 2928	17.4	11
46	Women Living with Familial Hypercholesterolemia: Challenges and Considerations Surrounding Their Care. *Current Atherosclerosis Reports*, **2020**, 22, 60	6	11
45	Genome-wide scan for circulating vascular adhesion protein-1 levels: MACROD2 as a potential transcriptional regulator of adipogenesis. *Journal of Diabetes Investigation*, **2018**, 9, 1067-1074	3.9	10
44	Validation of an Integrated Risk Tool, Including Polygenic Risk Score, for Atherosclerotic Cardiovascular Disease in Multiple Ethnicities and Ancestries. *American Journal of Cardiology*, **2021**, 148, 157-164	3	10
43	A case of complete heart block reverting to normal sinus rhythm after treatment for cardiac invasive Burkitt lymphoma. *Annals of Hematology*, **2007**, 86, 687-90	3	9
42	Usual Blood Pressure and New-Onset Diabetes Risk: Evidence From 4.1 Million Adults and a Meta-Analysis. *Journal of the American College of Cardiology*, **2016**, 67, 1656-1657	15.1	9
41	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). *American Journal of Cardiology*, **2016**, 118, 1275-1281	3	9
40	No evidence of a causal association of type 2 diabetes and glucose metabolism with atrial fibrillation. *Diabetologia*, **2019**, 62, 800-804	10.3	9
39	Short-Term Repeatability of Insulin Resistance Indexes in Older Adults: The Atherosclerosis Risk in Communities Study. *Journal of Clinical Endocrinology and Metabolism*, **2018**, 103, 2175-2181	5.6	8
38	PCSK9 inhibition: current concepts and lessons from human genetics. *Current Atherosclerosis Reports*, **2015**, 17, 487	6	8
37	How does morphology impact on diastolic function in hypertrophic cardiomyopathy? A single centre experience. *BMJ Open*, **2014**, 4, e004814	3	8
36	Latent obstruction and left atrial size are predictors of clinical deterioration leading to septal reduction in hypertrophic cardiomyopathy. *Journal of Cardiac Failure*, **2014**, 20, 236-43	3.3	8
35	Relationship between simple markers of insulin resistance and coronary artery calcification. *Journal of Clinical Lipidology*, **2017**, 11, 1007-1012	4.9	7
34	Predictive network modeling in human induced pluripotent stem cells identifies key driver genes for insulin responsiveness. *PLoS Computational Biology*, **2020**, 16, e1008491	5	7
33	The Clinical Genome Resource (ClinGen) Familial Hypercholesterolemia Variant Curation Expert Panel consensus guidelines for LDLR variant classification.. *Genetics in Medicine*, **2021**,	8.1	6
32	Cardiorespiratory Fitness, Body Mass Index, and Markers of Insulin Resistance in Apparently Healthy Women and Men. *American Journal of Medicine*, **2020**, 133, 825-830.e2	2.4	6

31	Statins Are Associated With Increased Insulin Resistance and Secretion. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2021**, 41, 2786-2797	9.4	6
30	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**, 62, 1185-1194	10.3	5
29	Isthmin-1 is an adipokine that promotes glucose uptake and improves glucose tolerance and hepatic steatosis. *Cell Metabolism*, **2021**, 33, 1836-1852.e11	24.6	5
28	Novel Therapies for Familial Hypercholesterolemia. *Current Treatment Options in Cardiovascular Medicine*, **2016**, 18, 64	2.1	4
27	The role of insulin as a key regulator of seeding, proliferation, and mRNA transcription of human pluripotent stem cells. *Stem Cell Research and Therapy*, **2019**, 10, 228	8.3	4
26	Exploring predisposition and treatment response--the promise of genomics. *Progress in Cardiovascular Diseases*, **2012**, 55, 56-63	8.5	4
25	Children with Heterozygous Familial Hypercholesterolemia in the United States: Data from the Cascade Screening for Awareness and Detection-FH Registry. *Journal of Pediatrics*, **2021**, 229, 70-77	3.6	4
24	Leveraging Human Genetics to Understand the Relation of LDL Cholesterol with Type 2 Diabetes. *Clinical Chemistry*, **2017**, 63, 1187-1189	5.5	3
23	CRISPR-Cas9-mediated knockout of SPRY2 in human hepatocytes leads to increased glucose uptake and lipid droplet accumulation. *BMC Endocrine Disorders*, **2019**, 19, 115	3.3	3
22	An integrated approach to identify environmental modulators of genetic risk factors for complex traits. *American Journal of Human Genetics*, **2021**, 108, 1866-1879	11	3
21	A guide for the diagnosis of rare and undiagnosed disease: beyond the exome.. *Genome Medicine*, **2022**, 14, 23	14.4	3
20	Large Q and S waves in lead III on the electrocardiogram distinguish patients with hypertrophic cardiomyopathy from athletes. *Heart*, **2018**, 104, 1871-1877	5.1	2
19	Cardiometabolic Effects of Glucagon-Like Peptide-1 Agonists. *Current Atherosclerosis Reports*, **2016**, 18, 7	6	2
18	Hyperuricaemia: the unintended consequence of insulin resistance/compensatory hyperinsulinaemia. Philanthropy gone awry. *Journal of Internal Medicine*, **2014**, 276, 196-8	10.8	2
17	Re: "Temporal relationship between uric acid concentration and risk of diabetes in a community-based study population". *American Journal of Epidemiology*, **2014**, 179, 1147-8	3.8	2
16	Coronary Artery Disease Risk of Familial Hypercholesterolemia Genetic Variants Independent of Clinically Observed Longitudinal Cholesterol Exposure.. *Circulation Genomic and Precision Medicine*, **2022**, CIRCGEN121003501	5.2	2
15	Genomic integrity of human induced pluripotent stem cells across nine studies in the NHLBI NextGen program. *Stem Cell Research*, **2020**, 46, 101803	1.6	2
14	Diverse Racial/Ethnic Group Underreporting and Underrepresentation in High-Impact Cholesterol Treatment Trials. *Circulation*, **2021**, 143, 2409-2411	16.7	2

13	Increasing Mortality Among Patients With Diabetes and Chronic Liver Disease From 2007 to 2017. *Clinical Gastroenterology and Hepatology*, **2020**, 18, 992-994	6.9	2
12	Genetics of Type 2 Diabetes: Opportunities for Precision Medicine: JACC Focus Seminar. *Journal of the American College of Cardiology*, **2021**, 78, 496-512	15.1	2
11	Signaling defects associated with insulin resistance in nondiabetic and diabetic individuals and modification by sex. *Journal of Clinical Investigation*, **2021**, 131,	15.9	2
10	Personalized Medicine and Cardiovascular Disease: From Genome to Bedside. *Current Cardiovascular Risk Reports*, **2011**, 5, 542-551	0.9	1
9	The Clinical Genome Resource (ClinGen) Familial Hypercholesterolemia Variant Curation Expert Panel consensus guidelines for LDLR variant classification		1
8	Generation of two heterozygous MYBPC3 mutation-carrying human iPSC lines, SCVIi001-A and SCVIi002-A, for modeling hypertrophic cardiomyopathy. *Stem Cell Research*, **2021**, 53, 102279	1.6	1
7	Health disparities in cardiometabolic risk among Black and Hispanic youth in the United States. *American Journal of Preventive Cardiology*, **2021**, 6, 100175	1.9	1
6	Online Patient Education Materials Related to Lipoprotein(a): Readability Assessment.. *Journal of Medical Internet Research*, **2022**, 24, e31284	7.6	0
5	The Human Arylamine N-Acetyltransferase Type 2 Gene: Genomics and Cardiometabolic Risk **2018**, 43-67		0
4	Integration of genetic colocalizations with physiological and pharmacological perturbations identifies cardiometabolic disease genes.. *Genome Medicine*, **2022**, 14, 31	14.4	0
3	Generation of two iPSC lines from hypertrophic cardiomyopathy patients carrying MYBPC3 and PRKAG2 variants.. *Stem Cell Research*, **2022**, 61, 102774	1.6	0
2	Familial Hypercholesterolemia. *Cardiac and Vascular Biology*, **2019**, 185-198	0.2	
1	Integration of Clinical Genetic Testing in Cardiovascular Care. *Current Genetic Medicine Reports*, **2016**, 4, 107-118	2.2	