

Joshua W Knowles

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	Coronary Artery Disease Risk of Familial Hypercholesterolemia Genetic Variants Independent of Clinically Observed Longitudinal Cholesterol Exposure.. <i>Circulation Genomic and Precision Medicine</i> , 2022 , CIRCGEN121003501	5.2	2
101	Online Patient Education Materials Related to Lipoprotein(a): Readability Assessment.. <i>Journal of Medical Internet Research</i> , 2022 , 24, e31284	7.6	0
100	A guide for the diagnosis of rare and undiagnosed disease: beyond the exome.. <i>Genome Medicine</i> , 2022 , 14, 23	14.4	3
99	Integration of genetic colocalizations with physiological and pharmacological perturbations identifies cardiometabolic disease genes.. <i>Genome Medicine</i> , 2022 , 14, 31	14.4	0
98	Generation of two iPSC lines from hypertrophic cardiomyopathy patients carrying MYBPC3 and PRKAG2 variants.. <i>Stem Cell Research</i> , 2022 , 61, 102774	1.6	0
97	The Clinical Genome Resource (ClinGen) Familial Hypercholesterolemia Variant Curation Expert Panel consensus guidelines for LDLR variant classification.. <i>Genetics in Medicine</i> , 2021 ,	8.1	6
96	Identification of rare and common regulatory variants in pluripotent cells using population-scale transcriptomics. <i>Nature Genetics</i> , 2021 , 53, 313-321	36.3	16
95	Generation of two heterozygous MYBPC3 mutation-carrying human iPSC lines, SCVII001-A and SCVII002-A, for modeling hypertrophic cardiomyopathy. <i>Stem Cell Research</i> , 2021 , 53, 102279	1.6	1
94	Health disparities in cardiometabolic risk among Black and Hispanic youth in the United States. <i>American Journal of Preventive Cardiology</i> , 2021 , 6, 100175	1.9	1
93	Diverse Racial/Ethnic Group Underreporting and Underrepresentation in High-Impact Cholesterol Treatment Trials. <i>Circulation</i> , 2021 , 143, 2409-2411	16.7	2
92	Validation of an Integrated Risk Tool, Including Polygenic Risk Score, for Atherosclerotic Cardiovascular Disease in Multiple Ethnicities and Ancestries. <i>American Journal of Cardiology</i> , 2021 , 148, 157-164	3	10
91	Children with Heterozygous Familial Hypercholesterolemia in the United States: Data From the Cascade Screening for Awareness and Detection-FH Registry. <i>Journal of Pediatrics</i> , 2021 , 229, 70-77	3.6	4
90	Genetics of Type 2 Diabetes: Opportunities for Precision Medicine: JACC Focus Seminar. <i>Journal of the American College of Cardiology</i> , 2021 , 78, 496-512	15.1	2
89	Statins Are Associated With Increased Insulin Resistance and Secretion. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 2021 , 41, 2786-2797	9.4	6
88	An integrated approach to identify environmental modulators of genetic risk factors for complex traits. <i>American Journal of Human Genetics</i> , 2021 , 108, 1866-1879	11	3
87	Signaling defects associated with insulin resistance in nondiabetic and diabetic individuals and modification by sex. <i>Journal of Clinical Investigation</i> , 2021 , 131,	15.9	2
86	Isthmin-1 is an adipokine that promotes glucose uptake and improves glucose tolerance and hepatic steatosis. <i>Cell Metabolism</i> , 2021 , 33, 1836-1852.e11	24.6	5

85	Discovery and quality analysis of a comprehensive set of structural variants and short tandem repeats. <i>Nature Communications</i> , 2020 , 11, 2928	17.4	11
84	FAM13A affects body fat distribution and adipocyte function. <i>Nature Communications</i> , 2020 , 11, 1465	17.4	17
83	Mitochondrial Dysfunction, Insulin Resistance, and Potential Genetic Implications. <i>Endocrinology</i> , 2020 , 161,	4.8	30
82	Association of insulin resistance, from mid-life to late-life, with aortic stiffness in late-life: the Atherosclerosis Risk in Communities Study. <i>Cardiovascular Diabetology</i> , 2020 , 19, 11	8.7	12
81	Predictive network modeling in human induced pluripotent stem cells identifies key driver genes for insulin responsiveness. <i>PLoS Computational Biology</i> , 2020 , 16, e1008491	5	7
80	Genomic integrity of human induced pluripotent stem cells across nine studies in the NHLBI NextGen program. <i>Stem Cell Research</i> , 2020 , 46, 101803	1.6	2
79	Cardiorespiratory Fitness, Body Mass Index, and Markers of Insulin Resistance in Apparently Healthy Women and Men. <i>American Journal of Medicine</i> , 2020 , 133, 825-830.e2	2.4	6
78	Women Living with Familial Hypercholesterolemia: Challenges and Considerations Surrounding Their Care. <i>Current Atherosclerosis Reports</i> , 2020 , 22, 60	6	11
77	Increasing Mortality Among Patients With Diabetes and Chronic Liver Disease From 2007 to 2017. <i>Clinical Gastroenterology and Hepatology</i> , 2020 , 18, 992-994	6.9	2
76	Longitudinal low density lipoprotein cholesterol goal achievement and cardiovascular outcomes among adult patients with familial hypercholesterolemia: The CASCADE FH registry. <i>Atherosclerosis</i> , 2019 , 289, 85-93	3.1	35
75	Genetic Testing and Risk Scores: Impact on Familial Hypercholesterolemia. <i>Frontiers in Cardiovascular Medicine</i> , 2019 , 6, 5	5.4	15
74	Impact of race/ethnicity on insulin resistance and hypertriglyceridaemia. <i>Diabetes and Vascular Disease Research</i> , 2019 , 16, 153-159	3.3	24
73	Trends in overall, cardiovascular and cancer-related mortality among individuals with diabetes reported on death certificates in the United States between 2007 and 2017. <i>Diabetologia</i> , 2019 , 62, 1185-1194 ⁵	10.3	5
72	Finding missed cases of familial hypercholesterolemia in health systems using machine learning. <i>Npj Digital Medicine</i> , 2019 , 2, 23	15.7	30
71	The role of insulin as a key regulator of seeding, proliferation, and mRNA transcription of human pluripotent stem cells. <i>Stem Cell Research and Therapy</i> , 2019 , 10, 228	8.3	4
70	Body composition and atrial fibrillation: a Mendelian randomization study. <i>European Heart Journal</i> , 2019 , 40, 1277-1282	9.5	23
69	CRISPR-Cas9-mediated knockout of SPRY2 in human hepatocytes leads to increased glucose uptake and lipid droplet accumulation. <i>BMC Endocrine Disorders</i> , 2019 , 19, 115	3.3	3
68	Precision screening for familial hypercholesterolaemia: a machine learning study applied to electronic health encounter data. <i>The Lancet Digital Health</i> , 2019 , 1, e393-e402	14.4	24

67	Familial Hypercholesterolemia. <i>Cardiac and Vascular Biology</i> , 2019 , 185-198	0.2	
66	No evidence of a causal association of type 2 diabetes and glucose metabolism with atrial fibrillation. <i>Diabetologia</i> , 2019 , 62, 800-804	10.3	9
65	Association of Statin Adherence With Mortality in Patients With Atherosclerotic Cardiovascular Disease. <i>JAMA Cardiology</i> , 2019 , 4, 206-213	16.2	108
64	Polygenic risk scores in coronary artery disease. <i>Current Opinion in Cardiology</i> , 2019 , 34, 435-440	2.1	18
63	Large Q and S waves in lead III on the electrocardiogram distinguish patients with hypertrophic cardiomyopathy from athletes. <i>Heart</i> , 2018 , 104, 1871-1877	5.1	2
62	Genome-wide scan for circulating vascular adhesion protein-1 levels: MACROD2 as a potential transcriptional regulator of adipogenesis. <i>Journal of Diabetes Investigation</i> , 2018 , 9, 1067-1074	3.9	10
61	First Trimester Plasma Glucose Values in Women without Diabetes are Associated with Risk for Congenital Heart Disease in Offspring. <i>Journal of Pediatrics</i> , 2018 , 195, 275-278	3.6	16
60	Clinical Genetic Testing for Familial Hypercholesterolemia: JACC Scientific Expert Panel. <i>Journal of the American College of Cardiology</i> , 2018 , 72, 662-680	15.1	215
59	Short-Term Repeatability of Insulin Resistance Indexes in Older Adults: The Atherosclerosis Risk in Communities Study. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2018 , 103, 2175-2181	5.6	8
58	Frequency of Statin Use in Patients With Low-Density Lipoprotein Cholesterol \geq 90 mg/dl from the Veterans Affairs Health System. <i>American Journal of Cardiology</i> , 2018 , 122, 756-761	3	17
57	Cardiovascular disease: The rise of the genetic risk score. <i>PLoS Medicine</i> , 2018 , 15, e1002546	11.6	81
56	The Human Arylamine N-Acetyltransferase Type 2 Gene: Genomics and Cardiometabolic Risk 2018 , 43-67		0
55	Large-Scale Phenome-Wide Association Study of Variants Demonstrates Protection Against Ischemic Stroke. <i>Circulation Genomic and Precision Medicine</i> , 2018 , 11, e002162	5.2	27
54	ClinVar database of global familial hypercholesterolemia-associated DNA variants. <i>Human Mutation</i> , 2018 , 39, 1631-1640	4.7	55
53	The role of registries and genetic databases in familial hypercholesterolemia. <i>Current Opinion in Lipidology</i> , 2017 , 28, 152-160	4.4	35
52	Access to Nonstatin Lipid-Lowering Therapies in Patients at High Risk of Atherosclerotic Cardiovascular Disease. <i>Circulation</i> , 2017 , 135, 2204-2206	16.7	28
51	Leveraging Human Genetics to Understand the Relation of LDL Cholesterol with Type 2 Diabetes. <i>Clinical Chemistry</i> , 2017 , 63, 1187-1189	5.5	3
50	Analysis of Transcriptional Variability in a Large Human iPSC Library Reveals Genetic and Non-genetic Determinants of Heterogeneity. <i>Cell Stem Cell</i> , 2017 , 20, 518-532.e9	18	164

49	Health disparities among adult patients with a phenotypic diagnosis of familial hypercholesterolemia in the CASCADE-FH patient registry. <i>Atherosclerosis</i> , 2017 , 267, 19-26	3.1	36
48	Induced Pluripotent Stem Cell-Derived Endothelial Cells in Insulin Resistance and Metabolic Syndrome. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 2017 , 37, 2038-2042	9.4	14
47	Cascade Screening for Familial Hypercholesterolemia and the Use of Genetic Testing. <i>JAMA - Journal of the American Medical Association</i> , 2017 , 318, 381-382	27.4	100
46	Relationship between simple markers of insulin resistance and coronary artery calcification. <i>Journal of Clinical Lipidology</i> , 2017 , 11, 1007-1012	4.9	7
45	Association Between Intensity of Statin Therapy and Mortality in Patients With Atherosclerotic Cardiovascular Disease. <i>JAMA Cardiology</i> , 2017 , 2, 47-54	16.2	92
44	Impact of a Genetic Risk Score for Coronary Artery Disease on Reducing Cardiovascular Risk: A Pilot Randomized Controlled Study. <i>Frontiers in Cardiovascular Medicine</i> , 2017 , 4, 53	5.4	29
43	Hypertriglyceridemia: A simple approach to identify insulin resistance and enhanced cardio-metabolic risk in patients with prediabetes. <i>Diabetes Research and Clinical Practice</i> , 2016 , 120, 156-61	7.4	12
42	US physician practices for diagnosing familial hypercholesterolemia: data from the CASCADE-FH registry. <i>Journal of Clinical Lipidology</i> , 2016 , 10, 1223-9	4.9	41
41	Novel Therapies for Familial Hypercholesterolemia. <i>Current Treatment Options in Cardiovascular Medicine</i> , 2016 , 18, 64	2.1	4
40	Impact of Septal Reduction on Left Atrial Size and Diastole in Hypertrophic Cardiomyopathy. <i>Echocardiography</i> , 2016 , 33, 686-94	1.5	17
39	Cardiometabolic Effects of Glucagon-Like Peptide-1 Agonists. <i>Current Atherosclerosis Reports</i> , 2016 , 18, 7	6	2
38	Treatment Gaps in Adults With Heterozygous Familial Hypercholesterolemia in the United States: Data From the CASCADE-FH Registry. <i>Circulation: Cardiovascular Genetics</i> , 2016 , 9, 240-9		126
37	Usual Blood Pressure and New-Onset Diabetes Risk: Evidence From 4.1 Million Adults and a Meta-Analysis. <i>Journal of the American College of Cardiology</i> , 2016 , 67, 1656-1657	15.1	9
36	Human induced pluripotent stem cell-derived cardiomyocytes recapitulate the predilection of breast cancer patients to doxorubicin-induced cardiotoxicity. <i>Nature Medicine</i> , 2016 , 22, 547-56	50.5	425
35	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). <i>American Journal of Cardiology</i> , 2016 , 118, 1275-1281	3	9
34	Integration of Clinical Genetic Testing in Cardiovascular Care. <i>Current Genetic Medicine Reports</i> , 2016 , 4, 107-118	2.2	
33	Use of high-intensity statins for patients with atherosclerotic cardiovascular disease in the Veterans Affairs Health System: Practice impact of the new cholesterol guidelines. <i>American Heart Journal</i> , 2016 , 182, 97-102	4.9	34
32	Nat1 Deficiency Is Associated with Mitochondrial Dysfunction and Exercise Intolerance in Mice. <i>Cell Reports</i> , 2016 , 17, 527-540	10.6	24

31	Prevalence and prognostic role of right ventricular involvement in stress-induced cardiomyopathy. <i>Journal of Cardiac Failure</i> , 2015 , 21, 419-425	3.3	22
30	Using Genetic Variants to Assess the Relationship Between Circulating Lipids and Type 2 Diabetes. <i>Diabetes</i> , 2015 , 64, 2676-84	0.9	83
29	Cardiopulmonary responses and prognosis in hypertrophic cardiomyopathy: a potential role for comprehensive noninvasive hemodynamic assessment. <i>JACC: Heart Failure</i> , 2015 , 3, 408-418	7.9	51
28	Maternal Midpregnancy Glucose Levels and Risk of Congenital Heart Disease in Offspring. <i>JAMA Pediatrics</i> , 2015 , 169, 1112-6	8.3	36
27	Identification and validation of N-acetyltransferase 2 as an insulin sensitivity gene. <i>Journal of Clinical Investigation</i> , 2015 , 125, 1739-51	15.9	67
26	The Agenda for Familial Hypercholesterolemia: A Scientific Statement From the American Heart Association. <i>Circulation</i> , 2015 , 132, 2167-92	16.7	377
25	PCSK9 inhibition: current concepts and lessons from human genetics. <i>Current Atherosclerosis Reports</i> , 2015 , 17, 487	6	8
24	Rationale and design of the familial hypercholesterolemia foundation CAsCade SCReening for Awareness and DEtection of Familial Hypercholesterolemia registry. <i>American Heart Journal</i> , 2014 , 167, 342-349.e17	4.9	63
23	Hyperuricaemia: the unintended consequence of insulin resistance/compensatory hyperinsulinaemia. Philanthropy gone awry. <i>Journal of Internal Medicine</i> , 2014 , 276, 196-8	10.8	2
22	Impact of type 2 diabetes susceptibility variants on quantitative glycemic traits reveals mechanistic heterogeneity. <i>Diabetes</i> , 2014 , 63, 2158-71	0.9	235
21	Simple, standardized incorporation of genetic risk into non-genetic risk prediction tools for complex traits: coronary heart disease as an example. <i>Frontiers in Genetics</i> , 2014 , 5, 254	4.5	39
20	How does morphology impact on diastolic function in hypertrophic cardiomyopathy? A single centre experience. <i>BMJ Open</i> , 2014 , 4, e004814	3	8
19	Genetic evidence for a normal-weight "metabolically obese" phenotype linking insulin resistance, hypertension, coronary artery disease, and type 2 diabetes. <i>Diabetes</i> , 2014 , 63, 4369-77	0.9	131
18	Re: "Temporal relationship between uric acid concentration and risk of diabetes in a community-based study population". <i>American Journal of Epidemiology</i> , 2014 , 179, 1147-8	3.8	2
17	Latent obstruction and left atrial size are predictors of clinical deterioration leading to septal reduction in hypertrophic cardiomyopathy. <i>Journal of Cardiac Failure</i> , 2014 , 20, 236-43	3.3	8
16	Prevalence and clinical correlates of right ventricular dysfunction in patients with hypertrophic cardiomyopathy. <i>American Journal of Cardiology</i> , 2014 , 113, 361-7	3	41
15	Measurement of insulin-mediated glucose uptake: direct comparison of the modified insulin suppression test and the euglycemic, hyperinsulinemic clamp. <i>Metabolism: Clinical and Experimental</i> , 2013 , 62, 548-53	12.7	42
14	Mendelian randomization studies do not support a causal role for reduced circulating adiponectin levels in insulin resistance and type 2 diabetes. <i>Diabetes</i> , 2013 , 62, 3589-98	0.9	95

13	Exploring predisposition and treatment response--the promise of genomics. <i>Progress in Cardiovascular Diseases</i> , 2012 , 55, 56-63	8.5	4
12	Randomized trial of personal genomics for preventive cardiology: design and challenges. <i>Circulation: Cardiovascular Genetics</i> , 2012 , 5, 368-76		23
11	Large-scale association analysis identifies 13 new susceptibility loci for coronary artery disease. <i>Nature Genetics</i> , 2011 , 43, 333-8	36.3	1394
10	Personalized Medicine and Cardiovascular Disease: From Genome to Bedside. <i>Current Cardiovascular Risk Reports</i> , 2011 , 5, 542-551	0.9	1
9	Phased whole-genome genetic risk in a family quartet using a major allele reference sequence. <i>PLoS Genetics</i> , 2011 , 7, e1002280	6	112
8	Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. <i>Nature Genetics</i> , 2010 , 42, 937-48	36.3	2267
7	Detailed physiologic characterization reveals diverse mechanisms for novel genetic Loci regulating glucose and insulin metabolism in humans. <i>Diabetes</i> , 2010 , 59, 1266-75	0.9	211
6	Clinical assessment incorporating a personal genome. <i>Lancet, The</i> , 2010 , 375, 1525-35	40	565
5	Failure to replicate an association of SNPs in the oxidized LDL receptor gene (OLR1) with CAD. <i>BMC Medical Genetics</i> , 2008 , 9, 23	2.1	24
4	A case of complete heart block reverting to normal sinus rhythm after treatment for cardiac invasive Burkitt lymphoma. <i>Annals of Hematology</i> , 2007 , 86, 687-90	3	9
3	Genetic susceptibility to peripheral arterial disease: a dark corner in vascular biology. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 2007 , 27, 2068-78	9.4	49
2	Association of polymorphisms in platelet and hemostasis system genes with acute myocardial infarction. <i>American Heart Journal</i> , 2007 , 154, 1052-8	4.9	43
1	The Clinical Genome Resource (ClinGen) Familial Hypercholesterolemia Variant Curation Expert Panel consensus guidelines for LDLR variant classification		1