Joshua W Knowles

List of Publications by Year
in descending order

111 papers	11,082 citations	76294 40 h-index	31818 101 g-index
117 all docs	117 docs citations	117 times ranked	19599 citing authors

#	Article	IF	Citations
1	Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. Nature Genetics, 2010, 42, 937-948.	9.4	2,634
2	Large-scale association analysis identifies 13 new susceptibility loci for coronary artery disease. Nature Genetics, 2011, 43, 333-338.	9.4	1,685
3	Clinical assessment incorporating a personal genome. Lancet, The, 2010, 375, 1525-1535.	6.3	637
4	Human induced pluripotent stem cell–derived cardiomyocytes recapitulate the predilection of breast cancer patients to doxorubicin-induced cardiotoxicity. Nature Medicine, 2016, 22, 547-556.	15.2	573
5	The Agenda for Familial Hypercholesterolemia. Circulation, 2015, 132, 2167-2192.	1.6	539
6	Clinical Genetic Testing for Familial Hypercholesterolemia. Journal of the American College of Cardiology, 2018, 72, 662-680.	1.2	387
7	Impact of Type 2 Diabetes Susceptibility Variants on Quantitative Glycemic Traits Reveals Mechanistic Heterogeneity. Diabetes, 2014, 63, 2158-2171.	0.3	297
8	Detailed Physiologic Characterization Reveals Diverse Mechanisms for Novel Genetic Loci Regulating Glucose and Insulin Metabolism in Humans. Diabetes, 2010, 59, 1266-1275.	0.3	237
9	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.	5.2	230
10	Association of Statin Adherence With Mortality in Patients With Atherosclerotic Cardiovascular Disease. JAMA Cardiology, 2019, 4, 206.	3.0	216
11	Genetic Evidence for a Normal-Weight "Metabolically Obese" Phenotype Linking Insulin Resistance, Hypertension, Coronary Artery Disease, and Type 2 Diabetes. Diabetes, 2014, 63, 4369-4377.	0.3	185
12	Treatment Gaps in Adults With Heterozygous Familial Hypercholesterolemia in the United States. Circulation: Cardiovascular Genetics, 2016, 9, 240-249.	5.1	170
13	Cascade Screening for Familial Hypercholesterolemia and the Use of Genetic Testing. JAMA - Journal of the American Medical Association, 2017, 318, 381.	3.8	138
14	Cardiovascular disease: The rise of the genetic risk score. PLoS Medicine, 2018, 15, e1002546.	3.9	138
15	Phased Whole-Genome Genetic Risk in a Family Quartet Using a Major Allele Reference Sequence. PLoS Genetics, 2011, 7, e1002280.	1.5	137
16	Association Between Intensity of Statin Therapy and Mortality in Patients With Atherosclerotic Cardiovascular Disease. JAMA Cardiology, 2017, 2, 47.	3.0	132
17	Mendelian Randomization Studies Do Not Support a Causal Role for Reduced Circulating Adiponectin Levels in Insulin Resistance and Type 2 Diabetes. Diabetes, 2013, 62, 3589-3598.	0.3	116
18	Using Genetic Variants to Assess the Relationship Between Circulating Lipids and Type 2 Diabetes. Diabetes, 2015, 64, 2676-2684.	0.3	114

#	Article	IF	Citations
19	A guide for the diagnosis of rare and undiagnosed disease: beyond the exome. Genome Medicine, 2022, 14, 23.	3.6	101
20	Mitochondrial Dysfunction, Insulin Resistance, and Potential Genetic Implications. Endocrinology, 2020, 161, .	1.4	96
21	Identification and validation of N-acetyltransferase 2 as an insulin sensitivity gene. Journal of Clinical Investigation, 2015, 125, 1739-1751.	3.9	94
22	ClinVar database of global familial hypercholesterolemiaâ€associated DNA variants. Human Mutation, 2018, 39, 1631-1640.	1.1	84
23	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.	1.2	76
24	Cardiopulmonary Responses and Prognosis in Hypertrophic Cardiomyopathy. JACC: Heart Failure, 2015, 3, 408-418.	1.9	72
25	Finding missed cases of familial hypercholesterolemia in health systems using machine learning. Npj Digital Medicine, 2019, 2, 23.	5.7	72
26	Health disparities among adult patients with a phenotypic diagnosis of familial hypercholesterolemia in the CASCADE-FHâ„¢ patient registry. Atherosclerosis, 2017, 267, 19-26.	0.4	64
27	Genetic Susceptibility to Peripheral Arterial Disease: A Dark Corner in Vascular Biology. Arteriosclerosis, Thrombosis, and Vascular Biology, 2007, 27, 2068-2078.	1.1	61
28	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.	0.4	60
29	US physician practices for diagnosing familial hypercholesterolemia: data from the CASCADE-FH registry. Journal of Clinical Lipidology, 2016, 10, 1223-1229.	0.6	57
30	Isthmin-1 is an adipokine that promotes glucose uptake and improves glucose tolerance and hepatic steatosis. Cell Metabolism, 2021, 33, 1836-1852.e11.	7.2	56
31	The Clinical Genome Resource (ClinGen) Familial Hypercholesterolemia Variant Curation Expert Panel consensus guidelines for LDLR variant classification. Genetics in Medicine, 2022, 24, 293-306.	1.1	53
32	Reducing the burden of disease and death from familial hypercholesterolemia: A call to action. American Heart Journal, 2014, 168, 807-811.	1.2	51
33	Precision screening for familial hypercholesterolaemia: a machine learning study applied to electronic health encounter data. The Lancet Digital Health, 2019, 1, e393-e402.	5.9	49
34	Statins Are Associated With Increased Insulin Resistance and Secretion. Arteriosclerosis, Thrombosis, and Vascular Biology, 2021, 41, 2786-2797.	1.1	49
35	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, 548-553.	1.5	48
36	Prevalence and Clinical Correlates of Right Ventricular Dysfunction in Patients With Hypertrophic Cardiomyopathy. American Journal of Cardiology, 2014, 113, 361-367.	0.7	48

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37	Large-Scale Phenome-Wide Association Study of *PCSK9* Variants Demonstrates Protection Against Ischemic Stroke. Circulation Genomic and Precision Medicine, 2018, 11, e002162.	1.6	48
38	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.	0.7	48
39	Body composition and atrial fibrillation: a Mendelian randomization study. European Heart Journal, 2019, 40, 1277-1282.	1.0	47
40	Impact of race/ethnicity on insulin resistance and hypertriglyceridaemia. Diabetes and Vascular Disease Research, 2019, 16, 153-159.	0.9	46
41	Association of polymorphisms in platelet and hemostasis system genes with acute myocardial infarction. American Heart Journal, 2007, 154, 1052-1058.	1.2	45
42	Maternal Midpregnancy Glucose Levels and Risk of Congenital Heart Disease in Offspring. JAMA Pediatrics, 2015, 169, 1112.	3.3	45
43	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.	1.1	44
44	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.	1.2	44
45	The role of registries and genetic databases in familial hypercholesterolemia. Current Opinion in Lipidology, 2017, 28, 152-160.	1.2	44
46	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.	1.1	44
47	Identification of rare and common regulatory variants in pluripotent cells using population-scale transcriptomics. Nature Genetics, 2021, 53, 313-321.	9.4	42
48	FAM13A affects body fat distribution and adipocyte function. Nature Communications, 2020, 11, 1465.	5.8	36
49	Nat1 Deficiency Is Associated with Mitochondrial Dysfunction and Exercise Intolerance in Mice. Cell Reports, 2016, 17, 527-540.	2.9	35
50	Access to Nonstatin Lipid-Lowering Therapies in Patients at High Risk of Atherosclerotic Cardiovascular Disease. Circulation, 2017, 135, 2204-2206.	1.6	34
51	Women Living with Familial Hypercholesterolemia: Challenges and Considerations Surrounding Their Care. Current Atherosclerosis Reports, 2020, 22, 60.	2.0	32
52	Polygenic risk scores in coronary artery disease. Current Opinion in Cardiology, 2019, 34, 435-440.	0.8	31
53	Failure to replicate an association of SNPs in the oxidized LDL receptor gene (OLR1) with CAD. BMC Medical Genetics, 2008, 9, 23.	2.1	29
54	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.	0.9	29

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55	Randomized Trial of Personal Genomics for Preventive Cardiology. Circulation: Cardiovascular Genetics, 2012, 5, 368-376.	5.1	28
56	Genetic Testing and Risk Scores: Impact on Familial Hypercholesterolemia. Frontiers in Cardiovascular Medicine, 2019, 6, 5.	1.1	28
57	Signaling defects associated with insulin resistance in nondiabetic and diabetic individuals and modification by sex. Journal of Clinical Investigation, 2021, 131, .	3.9	27
58	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.	2.7	24
59	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.	2.9	23
60	Prevalence and Prognostic Role of Right Ventricular Involvement in Stress-Induced Cardiomyopathy. Journal of Cardiac Failure, 2015, 21, 419-425.	0.7	22
61	Impact of Septal Reduction on Left Atrial Size and Diastole in Hypertrophic Cardiomyopathy. Echocardiography, 2016, 33, 686-694.	0.3	22
62	Discovery and quality analysis of a comprehensive set of structural variants and short tandem repeats. Nature Communications, 2020, 11, 2928.	5.8	22
63	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.	0.7	20
64	No evidence of a causal association of type 2 diabetes and glucose metabolism with atrial fibrillation. Diabetologia, 2019, 62, 800-804.	2.9	20
65	Induced Pluripotent Stem Cell–Derived Endothelial Cells in Insulin Resistance and Metabolic Syndrome. Arteriosclerosis, Thrombosis, and Vascular Biology, 2017, 37, 2038-2042.	1.1	19
66	Hypertriglyceridemia: A simple approach to identify insulin resistance and enhanced cardio-metabolic risk in patients with prediabetes. Diabetes Research and Clinical Practice, 2016, 120, 156-161.	1.1	18
67	Health disparities in cardiometabolic risk among Black and Hispanic youth in the United States. American Journal of Preventive Cardiology, 2021, 6, 100175.	1.3	18
68	Familial Hypercholesterolemia and the 2013 American College of Cardiology/American Heart Association Guidelines: Myths, Oversimplification, and Misinterpretation Versus Facts. American Journal of Cardiology, 2015, 116, 481-484.	0.7	16
69	How does morphology impact on diastolic function in hypertrophic cardiomyopathy? A single centre experience. BMJ Open, 2014, 4, e004814-e004814.	0.8	14
70	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.	0.6	14
71	Predictive network modeling in human induced pluripotent stem cells identifies key driver genes for insulin responsiveness. PLoS Computational Biology, 2020, 16, e1008491.	1.5	14
72	Metabolic Markers to Predict Incident Diabetes Mellitus in Statin-Treated Patients (from the Treating) Tj ETQq0 0 0 rgBT /Overlock 10 Tf American Journal of Cardiology, 2016, 118, 1275-1281.	0.7	13

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73	Genome‐wide scan for circulating vascular adhesion protein‐1 levels: <i><scp>MACROD</scp>2</i> as a potential transcriptional regulator of adipogenesis. Journal of Diabetes Investigation, 2018, 9, 1067-1074.	1.1	13
74	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.	0.9	13
75	Latent Obstruction and Left Atrial Size Are Predictors of Clinical Deterioration Leading to Septal Reduction in Hypertrophic Cardiomyopathy. Journal of Cardiac Failure, 2014, 20, 236-243.	0.7	12
76	Genetics of Type 2 Diabetes: Opportunities for Precision Medicine. Journal of the American College of Cardiology, 2021, 78, 496-512.	1.2	12
77	Usual Blood Pressure and New-Onset Diabetes Risk. Journal of the American College of Cardiology, 2016, 67, 1656-1657.	1.2	11
78	A case of complete heart block reverting to normal sinus rhythm after treatment for cardiac invasive Burkitt's lymphoma. Annals of Hematology, 2007, 86, 687-690.	0.8	10
79	Delisting <i>STAP1</i>. Arteriosclerosis, Thrombosis, and Vascular Biology, 2020, 40, 847-849.	1.1	10
80	Genomic integrity of human induced pluripotent stem cells across nine studies in the NHLBI NextGen program. Stem Cell Research, 2020, 46, 101803.	0.3	10
81	Online Patient Education Materials Related to Lipoprotein(a): Readability Assessment. Journal of Medical Internet Research, 2022, 24, e31284.	2.1	10
82	PCSK9 Inhibition: Current Concepts and Lessons from Human Genetics. Current Atherosclerosis Reports, 2015, 17, 487.	2.0	9
83	Relationship between simple markers of insulin resistance and coronary artery calcification. Journal of Clinical Lipidology, 2017, 11, 1007-1012.	0.6	9
84	An integrated approach to identify environmental modulators of genetic risk factors for complex traits. American Journal of Human Genetics, 2021, 108, 1866-1879.	2.6	9
85	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.	1.8	8
86	Diverse Racial/Ethnic Group Underreporting and Underrepresentation in High-Impact Cholesterol Treatment Trials. Circulation, 2021, 143, 2409-2411.	1.6	8
87	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.	2.4	7
88	Interactions of physical activity, muscular fitness, adiposity, and genetic risk for NAFLD. Hepatology Communications, 2022, 6, 1516-1526.	2.0	7
89	Integration of genetic colocalizations with physiological and pharmacological perturbations identifies cardiometabolic disease genes. Genome Medicine, 2022, 14, 31.	3.6	7
90	Enough Evidence, Time to Act!. Circulation, 2016, 134, 20-23.	1.6	6

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91	Novel Therapies for Familial Hypercholesterolemia. Current Treatment Options in Cardiovascular Medicine, 2016, 18, 64.	0.4	6
92	CRISPR-Cas9-mediated knockout of SPRY2 in human hepatocytes leads to increased glucose uptake and lipid droplet accumulation. BMC Endocrine Disorders, 2019, 19, 115.	0.9	6
93	Increasing Mortality Among Patients With Diabetes and Chronic Liver Disease From 2007 to 2017. Clinical Gastroenterology and Hepatology, 2020, 18, 992-994.	2.4	6
94	Coronary Artery Disease Risk of Familial Hypercholesterolemia Genetic Variants Independent of Clinically Observed Longitudinal Cholesterol Exposure. Circulation Genomic and Precision Medicine, 2022, 15, CIRCGEN121003501.	1.6	6
95	Statins in Familial Hypercholesterolemia. Journal of the American College of Cardiology, 2016, 68, 261-264.	1.2	5
96	Large Q and S waves in lead III on the electrocardiogram distinguish patients with hypertrophic cardiomyopathy from athletes. Heart, 2018, 104, 1871-1877.	1.2	5
97	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.	0.3	5
98	Exploring Predisposition and Treatment Response—the Promise of Genomics. Progress in Cardiovascular Diseases, 2012, 55, 56-63.	1.6	4
99	Leveraging Human Genetics to Understand the Relation of LDL Cholesterol with Type 2 Diabetes. Clinical Chemistry, 2017, 63, 1187-1189.	1.5	4
100	Generation of two iPSC lines from hypertrophic cardiomyopathy patients carrying MYBPC3 and PRKAG2 variants. Stem Cell Research, 2022, 61, 102774.	0.3	4
101	Standards of Evidence and Mechanistic Inference in Autosomal Recessive Hypercholesterolemia. Arteriosclerosis, Thrombosis, and Vascular Biology, 2016, 36, 1465-1466.	1.1	3
102	Re: "Temporal Relationship Between Uric Acid Concentration and Risk of Diabetes in a Community-Based Study Population". American Journal of Epidemiology, 2014, 179, 1147-1148.	1.6	2
103	Hyperuricaemia: the unintended consequence of insulin resistance/compensatory hyperinsulinaemia. Philanthropy gone awry. Journal of Internal Medicine, 2014, 276, 196-198.	2.7	2
104	Cardiometabolic Effects of Glucagon-Like Peptide-1 Agonists. Current Atherosclerosis Reports, 2016, 18, 7.	2.0	2
105	The Human Arylamine <i>N</i>-Acetyltransferase Type 2 Gene: Genomics and Cardiometabolic Risk. , 2018, , 43-67.		2
106	Personalized Medicine and Cardiovascular Disease: From Genome to Bedside. Current Cardiovascular Risk Reports, 2011, 5, 542-551.	0.8	1
107	Is ACS in Young Patients a "Canary in the Coal Mine" for Familial Hypercholesterolemia?. Journal of the American College of Cardiology, 2017, 70, 1741-1743.	1.2	1
108	Abstract 12169: LDL-C Levels and Treatment Patterns Among Adults With Heterozygous Familial Hypercholesterolemia in the United States: Data From the CASCADE-FH Registry. Circulation, 2015, 132, .	1.6	1

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109	Integration of Clinical Genetic Testing in Cardiovascular Care. Current Genetic Medicine Reports, 2016, 4, 107-118.	1.9	0
110	Abstract 16184: Elevated Maternal Glucose Levels During Gestation may Confer Specific Risk for Tetralogy of Fallot. Circulation, 2015, 132, .	1.6	0
111	Familial Hypercholesterolemia. Cardiac and Vascular Biology, 2019, , 185-198.	0.2	0