

James S Ware

List of Publications by Citations

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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.

119
papers

14,765
citations

38
h-index

121
g-index

142
ext. papers

21,138
ext. citations

11.9
avg, IF

5.55
L-index

#	Paper	IF	Citations
119	Analysis of protein-coding genetic variation in 60,706 humans. <i>Nature</i> , 2016 , 536, 285-91	50.4	6940
118	The mutational constraint spectrum quantified from variation in 141,456 humans. <i>Nature</i> , 2020 , 581, 434-443	50.4	2278
117	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. <i>Science</i> , 2015 , 350, 1262-6	33.3	406
116	Reassessment of Mendelian gene pathogenicity using 7,855 cardiomyopathy cases and 60,706 reference samples. <i>Genetics in Medicine</i> , 2017 , 19, 192-203	8.1	386
115	The mutational constraint spectrum quantified from variation in 141,456 humans		381
114	Shared Genetic Predisposition in Peripartum and Dilated Cardiomyopathies. <i>New England Journal of Medicine</i> , 2016 , 374, 233-41	59.2	290
113	Integrated allelic, transcriptional, and phenomic dissection of the cardiac effects of titin truncations in health and disease. <i>Science Translational Medicine</i> , 2015 , 7, 270ra6	17.5	267
112	Genotype and Lifetime Burden of Disease in Hypertrophic Cardiomyopathy: Insights from the Sarcomeric Human Cardiomyopathy Registry (SHaRe). <i>Circulation</i> , 2018 , 138, 1387-1398	16.7	210
111	Using high-resolution variant frequencies to empower clinical genome interpretation. <i>Genetics in Medicine</i> , 2017 , 19, 1151-1158	8.1	208
110	Withdrawal of pharmacological treatment for heart failure in patients with recovered dilated cardiomyopathy (TRED-HF): an open-label, pilot, randomised trial. <i>Lancet, The</i> , 2019 , 393, 61-73	40	198
109	Titin-truncating variants affect heart function in disease cohorts and the general population. <i>Nature Genetics</i> , 2017 , 49, 46-53	36.3	179
108	Adaptation and validation of the ACMG/AMP variant classification framework for MYH7-associated inherited cardiomyopathies: recommendations by ClinGen's Inherited Cardiomyopathy Expert Panel. <i>Genetics in Medicine</i> , 2018 , 20, 351-359	8.1	173
107	Reappraisal of Reported Genes for Sudden Arrhythmic Death: Evidence-Based Evaluation of Gene Validity for Brugada Syndrome. <i>Circulation</i> , 2018 , 138, 1195-1205	16.7	158
106	Evaluating the Clinical Validity of Hypertrophic Cardiomyopathy Genes. <i>Circulation Genomic and Precision Medicine</i> , 2019 , 12, e002460	5.2	132
105	Utility of Post-Mortem Genetic Testing in Cases of Sudden Arrhythmic Death Syndrome. <i>Journal of the American College of Cardiology</i> , 2017 , 69, 2134-2145	15.1	126
104	Endonuclease G is a novel determinant of cardiac hypertrophy and mitochondrial function. <i>Nature</i> , 2011 , 478, 114-8	50.4	114
103	Genetic Etiology for Alcohol-Induced Cardiac Toxicity. <i>Journal of the American College of Cardiology</i> , 2018 , 71, 2293-2302	15.1	112

102	Genetic Variants Associated With Cancer Therapy-Induced Cardiomyopathy. <i>Circulation</i> , 2019 , 140, 31-41	16.7	110
101	Defining the genetic architecture of hypertrophic cardiomyopathy: re-evaluating the role of non-sarcomeric genes. <i>European Heart Journal</i> , 2017 , 38, 3461-3468	9.5	101
100	An International, Multicentered, Evidence-Based Reappraisal of Genes Reported to Cause Congenital Long QT Syndrome. <i>Circulation</i> , 2020 , 141, 418-428	16.7	95
99	Effects of myosin variants on interacting-heads motif explain distinct hypertrophic and dilated cardiomyopathy phenotypes. <i>ELife</i> , 2017 , 6,	8.9	87
98	Reevaluating the Genetic Contribution of Monogenic Dilated Cardiomyopathy. <i>Circulation</i> , 2020 , 141, 387-398	16.7	71
97	Sudden Death and Left Ventricular Involvement in Arrhythmogenic Cardiomyopathy. <i>Circulation</i> , 2019 , 139, 1786-1797	16.7	70
96	Role of titin in cardiomyopathy: from DNA variants to patient stratification. <i>Nature Reviews Cardiology</i> , 2018 , 15, 241-252	14.8	69
95	Development of a Comprehensive Sequencing Assay for Inherited Cardiac Condition Genes. <i>Journal of Cardiovascular Translational Research</i> , 2016 , 9, 3-11	3.3	63
94	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. <i>Annals of Neurology</i> , 2018 , 83, 1105-1124	9.4	59
93	Phenotype and Clinical Outcomes of Titin Cardiomyopathy. <i>Journal of the American College of Cardiology</i> , 2017 , 70, 2264-2274	15.1	57
92	Interpreting de novo Variation in Human Disease Using denovolyzeR. <i>Current Protocols in Human Genetics</i> , 2015 , 87, 7.25.1-7.25.15	3.2	56
91	Quantitative approaches to variant classification increase the yield and precision of genetic testing in Mendelian diseases: the case of hypertrophic cardiomyopathy. <i>Genome Medicine</i> , 2019 , 11, 5	14.4	54
90	Dilated Cardiomyopathy Due to BCL2-Associated Atrogin-1 (BAG3) Mutations. <i>Journal of the American College of Cardiology</i> , 2018 , 72, 2471-2481	15.1	53
89	Towards clinical molecular diagnosis of inherited cardiac conditions: a comparison of bench-top genome DNA sequencers. <i>PLoS ONE</i> , 2013 , 8, e67744	3.7	48
88	The effect of LRRK2 loss-of-function variants in humans. <i>Nature Medicine</i> , 2020 , 26, 869-877	50.5	47
87	CardioClassifier: disease- and gene-specific computational decision support for clinical genome interpretation. <i>Genetics in Medicine</i> , 2018 , 20, 1246-1254	8.1	45
86	Hypertrophic Cardiomyopathy With Left Ventricular Systolic Dysfunction: Insights From the SHaRe Registry. <i>Circulation</i> , 2020 , 141, 1371-1383	16.7	43
85	Truncating Variants in Titin Independently Predict Early Arrhythmias in Patients With Dilated Cardiomyopathy. <i>Journal of the American College of Cardiology</i> , 2017 , 69, 2466-2468	15.1	40

84	Natural genetic variation of the cardiac transcriptome in non-diseased donors and patients with dilated cardiomyopathy. <i>Genome Biology</i> , 2017 , 18, 170	18.3	40
83	Next generation sequencing for clinical diagnostics and personalised medicine: implications for the next generation cardiologist. <i>Heart</i> , 2012 , 98, 276-81	5.1	39
82	Detection of mutations in KLHL3 and CUL3 in families with FHHT (familial hyperkalaemic hypertension or Gordon's syndrome). <i>Clinical Science</i> , 2014 , 126, 721-6	6.5	38
81	Characterising the loss-of-function impact of 5' untranslated region variants in 15,708 individuals. <i>Nature Communications</i> , 2020 , 11, 2523	17.4	35
80	Phenotypic and pharmacogenetic evaluation of patients with thiazide-induced hyponatremia. <i>Journal of Clinical Investigation</i> , 2017 , 127, 3367-3374	15.9	35
79	Shared genetic pathways contribute to risk of hypertrophic and dilated cardiomyopathies with opposite directions of effect. <i>Nature Genetics</i> , 2021 , 53, 128-134	36.3	35
78	Evidence-Based Assessment of Genes in Dilated Cardiomyopathy. <i>Circulation</i> , 2021 , 144, 7-19	16.7	34
77	Loss of RNA expression and allele-specific expression associated with congenital heart disease. <i>Nature Communications</i> , 2016 , 7, 12824	17.4	33
76	Common genetic variants and modifiable risk factors underpin hypertrophic cardiomyopathy susceptibility and expressivity. <i>Nature Genetics</i> , 2021 , 53, 135-142	36.3	33
75	Paralogue annotation identifies novel pathogenic variants in patients with Brugada syndrome and catecholaminergic polymorphic ventricular tachycardia. <i>Journal of Medical Genetics</i> , 2014 , 51, 35-44	5.8	31
74	Quality assurance of item writing: during the introduction of multiple choice questions in medicine for high stakes examinations. <i>Medical Teacher</i> , 2009 , 31, 238-43	3	31
73	Next generation diagnostics in inherited arrhythmia syndromes : a comparison of two approaches. <i>Journal of Cardiovascular Translational Research</i> , 2013 , 6, 94-103	3.3	28
72	Moderate Physical Activity in Healthy Adults Is Associated With Cardiac Remodeling. <i>Circulation: Cardiovascular Imaging</i> , 2016 , 9,	3.9	27
71	Paralogous annotation of disease-causing variants in long QT syndrome genes. <i>Human Mutation</i> , 2012 , 33, 1188-1191	4.7	26
70	Genetic and functional insights into the fractal structure of the heart. <i>Nature</i> , 2020 , 584, 589-594	50.4	26
69	Analysis of 51 proposed hypertrophic cardiomyopathy genes from genome sequencing data in sarcomere negative cases has negligible diagnostic yield. <i>Genetics in Medicine</i> , 2019 , 21, 1576-1584	8.1	25
68	Role of Targeted Therapy in Dilated Cardiomyopathy: The Challenging Road Toward a Personalized Approach. <i>Journal of the American Heart Association</i> , 2019 , 8, e012514	6	21
67	Recovery of Cardiac Function in Cardiomyopathy Caused by Titin Truncation. <i>JAMA Cardiology</i> , 2016 , 1, 234-5	16.2	21

66	Identification of an I-dependent and I-mediated proarrhythmic mechanism in cardiomyocytes derived from pluripotent stem cells of a Brugada syndrome patient. <i>Scientific Reports</i> , 2018 , 8, 11246	4.9	20
65	Association of Titin-Truncating Genetic Variants With Life-threatening Cardiac Arrhythmias in Patients With Dilated Cardiomyopathy and Implanted Defibrillators. <i>JAMA Network Open</i> , 2019 , 2, e196520	10.4	20
64	Clinical characteristics and outcomes in childhood-onset hypertrophic cardiomyopathy. <i>European Heart Journal</i> , 2021 , 42, 1988-1996	9.5	20
63	Bayesian models for syndrome- and gene-specific probabilities of novel variant pathogenicity. <i>Genome Medicine</i> , 2015 , 7, 5	14.4	19
62	Spatial and Functional Distribution of Pathogenic Variants and Clinical Outcomes in Patients With Hypertrophic Cardiomyopathy. <i>Circulation Genomic and Precision Medicine</i> , 2020 , 13, 396-405	5.2	19
61	Titin truncating mutations: A rare cause of dilated cardiomyopathy in the young. <i>Progress in Pediatric Cardiology</i> , 2016 , 40, 41-45	0.4	17
60	Ajmaline blocks I and I without eliciting differences between Brugada syndrome patient and control human pluripotent stem cell-derived cardiac clusters. <i>Stem Cell Research</i> , 2017 , 25, 233-244	1.6	17
59	Clinical Features and Natural History of PRKAG2 Variant Cardiac Glycogenosis. <i>Journal of the American College of Cardiology</i> , 2020 , 76, 186-197	15.1	16
58	ClinVar data parsing. <i>Wellcome Open Research</i> , 2017 , 2, 33	4.8	16
57	Genomic variant sharing: a position statement. <i>Wellcome Open Research</i> , 2019 , 4, 22	4.8	16
56	Temporal Trend of Age at Diagnosis in Hypertrophic Cardiomyopathy: An Analysis of the International Sarcomeric Human Cardiomyopathy Registry. <i>Circulation: Heart Failure</i> , 2020 , 13, e007230	7.6	16
55	The yield of postmortem genetic testing in sudden death cases with structural findings at autopsy. <i>European Journal of Human Genetics</i> , 2020 , 28, 17-22	5.3	16
54	Histiocytoid cardiomyopathy and microphthalmia with linear skin defects syndrome: phenotypes linked by truncating variants in. <i>Journal of Physical Education and Sports Management</i> , 2017 , 3, a001271	2.8	15
53	Gene family information facilitates variant interpretation and identification of disease-associated genes in neurodevelopmental disorders. <i>Genome Medicine</i> , 2020 , 12, 28	14.4	13
52	Predictors of left ventricular remodelling in patients with dilated cardiomyopathy - a cardiovascular magnetic resonance study. <i>European Journal of Heart Failure</i> , 2020 , 22, 1160-1170	12.3	13
51	Enhancing rare variant interpretation in inherited arrhythmias through quantitative analysis of consortium disease cohorts and population controls. <i>Genetics in Medicine</i> , 2021 , 23, 47-58	8.1	13
50	Systematic large-scale assessment of the genetic architecture of left ventricular noncompaction reveals diverse etiologies. <i>Genetics in Medicine</i> , 2021 , 23, 856-864	8.1	12
49	Associations Between Female Sex, Sarcomere Variants, and Clinical Outcomes in Hypertrophic Cardiomyopathy. <i>Circulation Genomic and Precision Medicine</i> , 2021 , 14, e003062	5.2	10

48	Diagnostic yield of hypertrophic cardiomyopathy in first-degree relatives of decedents with idiopathic left ventricular hypertrophy. <i>Europace</i> , 2020 , 22, 632-642	3.9	10
47	ZBTB17 (MIZ1) Is Important for the Cardiac Stress Response and a Novel Candidate Gene for Cardiomyopathy and Heart Failure. <i>Circulation: Cardiovascular Genetics</i> , 2015 , 8, 643-52		9
46	Integrative genomics in cardiovascular medicine. <i>Cardiovascular Research</i> , 2013 , 97, 623-30	9.9	8
45	Titin Circular RNAs Create a Back-Splice Motif Essential for SRSF10 Splicing. <i>Circulation</i> , 2021 , 143, 1502-1512	16.7	8
44	Using High-Resolution Variant Frequencies Empowers Clinical Genome Interpretation and Enables Investigation of Genetic Architecture. <i>American Journal of Human Genetics</i> , 2019 , 104, 187-190	11	8
43	Annotating high-impact 5'untranslated region variants with the UTRannotator. <i>Bioinformatics</i> , 2021 , 37, 1171-1173	7.2	8
42	Human loss-of-function variants suggest that partial LRRK2 inhibition is a safe therapeutic strategy for Parkinson disease		7
41	Gene family information facilitates variant interpretation and identification of disease-associated genes		6
40	Understanding the genetics of adult-onset dilated cardiomyopathy: what a clinician needs to know. <i>European Heart Journal</i> , 2021 , 42, 2384-2396	9.5	6
39	Worldwide differences in primary prevention implantable cardioverter defibrillator utilization and outcomes in hypertrophic cardiomyopathy. <i>European Heart Journal</i> , 2021 , 42, 3932-3944	9.5	6
38	Next-generation sequencing using microfluidic PCR enrichment for molecular autopsy. <i>BMC Cardiovascular Disorders</i> , 2019 , 19, 174	2.3	5
37	Genomic variant sharing: a position statement. <i>Wellcome Open Research</i> , 4 , 22	4.8	5
36	Reassessment of Mendelian gene pathogenicity using 7,855 cardiomyopathy cases and 60,706 reference samples		5
35	Characterising the loss-of-function impact of 5'untranslated region variants in whole genome sequence data from 15,708 individuals		5
34	Evaluation of gene validity for CPVT and short QT syndrome in sudden arrhythmic death. <i>European Heart Journal</i> , 2021 ,	9.5	5
33	An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. <i>Human Mutation</i> , 2020 , 41, 1577-1587	4.7	4
32	Using high-resolution variant frequencies to empower clinical genome interpretation		4
31	Genetic Studies of Hypertrophic Cardiomyopathy in Singaporeans Identify Variants in and That Are Common in Chinese Patients. <i>Circulation Genomic and Precision Medicine</i> , 2020 , 13, 424-434	5.2	4

30	Disease-specific variant pathogenicity prediction significantly improves variant interpretation in inherited cardiac conditions. <i>Genetics in Medicine</i> , 2021 , 23, 69-79	8.1	4
29	The Gene Curation Coalition: A global effort to harmonize gene-disease evidence resources.. <i>Genetics in Medicine</i> , 2022 ,	8.1	4
28	Improving the Understanding of Genetic Variants in Rare Disease With Large-scale Reference Populations. <i>JAMA - Journal of the American Medical Association</i> , 2019 , 322, 1305-1306	27.4	3
27	Founder Mutation in N Terminus of Cardiac Troponin I Causes Malignant Hypertrophic Cardiomyopathy. <i>Circulation Genomic and Precision Medicine</i> , 2020 , 13, 444-452	5.2	3
26	Computational prediction of protein subdomain stability in MYBPC3 enables clinical risk stratification in hypertrophic cardiomyopathy and enhances variant interpretation. <i>Genetics in Medicine</i> , 2021 , 23, 1281-1287	8.1	3
25	Clinical impact of re-evaluating genes and variants implicated in dilated cardiomyopathy. <i>Genetics in Medicine</i> , 2021 , 23, 2186-2193	8.1	3
24	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. <i>Nature</i> , 2021 , 597, E3-E4	50.4	3
23	Phenotypic Expression and Outcomes in Individuals With Rare Genetic Variants of Hypertrophic Cardiomyopathy. <i>Journal of the American College of Cardiology</i> , 2021 , 78, 1097-1110	15.1	3
22	Response by Ho et al to Letter Regarding Article, "Genotype and Lifetime Burden of Disease in Hypertrophic Cardiomyopathy: Insights From the Sarcomeric Human Cardiomyopathy Registry (SHaRe)". <i>Circulation</i> , 2019 , 139, 1559-1560	16.7	2
21	NECTAR: a database of codon-centric missense variant annotations. <i>Nucleic Acids Research</i> , 2014 , 42, D1013-9	20.1	2
20	Republished review: Next generation sequencing for clinical diagnostics and personalised medicine: implications for the next generation cardiologist. <i>Postgraduate Medical Journal</i> , 2012 , 88, 234-9	2	2
19	Analysis of HCM in an understudied population reveals a new mechanism of pathogenicity		2
18	The Egyptian Collaborative Cardiac Genomics (ECCO-GEN) Project: defining a healthy volunteer cohort. <i>Npj Genomic Medicine</i> , 2020 , 5, 46	6.2	2
17	Moderate excess alcohol consumption and adverse cardiac remodelling in dilated cardiomyopathy. <i>Heart</i> , 2021 ,	5.1	2
16	Precision Phenotyping of Dilated Cardiomyopathy Using Multidimensional Data. <i>Journal of the American College of Cardiology</i> , 2022 , 79, 2219-2232	15.1	2
15	50 Incremental diagnostic value of cardiovascular magnetic resonance in young adult survivors of sudden cardiac arrest. <i>Heart</i> , 2017 , 103, A39-A39	5.1	1
14	Exposure to Elevated Nitrogen Dioxide Concentrations and Cardiac Remodeling in Patients With Dilated Cardiomyopathy.. <i>Journal of Cardiac Failure</i> , 2022 ,	3.3	1
13	Quantitative approaches to variant classification increase the yield and precision of genetic testing in Mendelian diseases: The case of hypertrophic cardiomyopathy		1

12	Genomics of Egyptian Healthy Volunteers: The EHVol Study			1
11	The genetic architecture of left ventricular non-compaction reveals both substantial overlap with other cardiomyopathies and a distinct aetiology in a subset of cases			1
10	Harmonizing the Collection of Clinical Data on Genetic Testing Requisition Forms to Enhance Variant Interpretation in Hypertrophic Cardiomyopathy (HCM): A Study from the ClinGen Cardiomyopathy Variant Curation Expert Panel. <i>Journal of Molecular Diagnostics</i> , 2021 , 23, 589-598	5.1		1
9	Genetic and environmental determinants of diastolic heart function			1
8	New Variant With a Previously Unrecognized Mechanism of Pathogenicity in Hypertrophic Cardiomyopathy. <i>Circulation</i> , 2021 , 144, 754-757	16.7		1
7	Outcomes and phenotypic expression of rare variants in hypertrophic cardiomyopathy genes amongst UK Biobank participants			1
6	Correspondence on "ACMG SF v3.0 list for reporting of secondary findings in clinical exome and genome sequencing: a policy statement of the American College of Medical Genetics and Genomics (ACMG)" by Miller et al.. <i>Genetics in Medicine</i> , 2021 ,	8.1		0
5	Prognostic Significance of Nonischemic Myocardial Fibrosis in Patients With Normal LV Volumes and Ejection-Fraction. <i>JACC: Cardiovascular Imaging</i> , 2021 , 14, 2353-2365	8.4		0
4	Genetic and environmental determinants of diastolic heart function. 2022 , 1, 361-371			0
3	127 Relationship between plasma concentrations of b-type natriuretic peptide and exercise capacity in hypertrophic cardiomyopathy. <i>Heart</i> , 2017 , 103, A96-A97	5.1		
2	Effect of taurine administration on symptoms, severity, or clinical outcome of dilated cardiomyopathy and heart failure in humans: a systematic review. <i>Wellcome Open Research</i> , 7, 9	4.8		
1	Effect of taurine administration on symptoms, severity, or clinical outcome of dilated cardiomyopathy and heart failure in humans: a systematic review. <i>Wellcome Open Research</i> , 7, 9	4.8		