James S Ware

List of Publications by Year
in descending order

117 papers	25,543 citations	50170 46 h-index	24179 110 g-index
142 all docs	142 docs citations	142 times ranked	44342 citing authors

#	Article	IF	Citations
1	Moderate excess alcohol consumption and adverse cardiac remodelling in dilated cardiomyopathy. Heart, 2022, 108, 619-625.	1.2	6
2	Evaluation of gene validity for CPVT and short QT syndrome in sudden arrhythmic death. European Heart Journal, 2022, 43, 1500-1510.	1.0	57
3	Quantifying prediction of pathogenicity for within-codon concordance (PM5) using 7541 functional classifications of BRCA1 and MSH2 missense variants. Genetics in Medicine, 2022, 24, 552-563.	1.1	5
4	Correspondence on â€œACMG 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)â€•byÂMiller etÂal. Genetics in Medicine, 2022, 24, 744-746.	1.1	17
5	Quantifying evidence toward pathogenicity for rare phenotypes: The case of succinate dehydrogenase genes, SDHB and SDHD. Genetics in Medicine, 2022, 24, 41-50.	1.1	5
6	Direct and indirect effect of the COVID-19 pandemic on patients with cardiomyopathy. Open Heart, 2022, 9, e001918.	0.9	3
7	Exposure to Elevated Nitrogen Dioxide Concentrations and Cardiac Remodeling in Patients With Dilated Cardiomyopathy. Journal of Cardiac Failure, 2022, 28, 924-934.	0.7	6
8	European Heart Rhythm Association (EHRA)/Heart Rhythm Society (HRS)/Asia Pacific Heart Rhythm Society (APHRS)/Latin American Heart Rhythm Society (LAHRS) Expert Consensus Statement on the state of genetic testing for cardiac diseases. Europace, 2022, 24, 1307-1367.	0.7	108
9	European Heart Rhythm Association (EHRA)/Heart Rhythm Society (HRS)/Asia Pacific Heart Rhythm Society (APHRS)/Latin American Heart Rhythm Society (LAHRS) Expert Consensus Statement on the State of Genetic Testing for Cardiac Diseases. Heart Rhythm, 2022, 19, e1-e60.	0.3	78
10	European Heart Rhythm Association (<scp>EHRA</scp>)/Heart Rhythm Society (<scp>HRS</scp>)/Asia Pacific Heart Rhythm Society (<scp>APHRS</scp>)/Latin American Heart Rhythm Society (<scp>LAHRS</scp>) Expert Consensus Statement on the state of genetic testing for cardiac diseases. Journal of Arrhythmia, 2022, 38, 491-553.	0.5	24
11	Genetic and environmental determinants of diastolic heart function. , 2022, 1, 361-371.		12
12	The Gene Curation Coalition: A global effort to harmonize geneâ€“disease evidence resources. Genetics in Medicine, 2022, 24, 1732-1742.	1.1	56
13	Precision Phenotyping of Dilated Cardiomyopathy Using Multidimensional Data. Journal of the American College of Cardiology, 2022, 79, 2219-2232.	1.2	24
14	Associations Between Female Sex, Sarcomere Variants, and Clinical Outcomes in Hypertrophic Cardiomyopathy. Circulation Genomic and Precision Medicine, 2021, 14, e003062.	1.6	38
15	Annotating high-impact 5â€²untranslated region variants with the UTRannotator. Bioinformatics, 2021, 37, 1171-1173.	1.8	27
16	Disease-specific variant pathogenicity prediction significantly improves variant interpretation in inherited cardiac conditions. Genetics in Medicine, 2021, 23, 69-79.	1.1	39
17	Enhancing rare variant interpretation in inherited arrhythmias through quantitative analysis of consortium disease cohorts and population controls. Genetics in Medicine, 2021, 23, 47-58.	1.1	57
18	Common genetic variants and modifiable risk factors underpin hypertrophic cardiomyopathy susceptibility and expressivity. Nature Genetics, 2021, 53, 135-142.	9.4	165

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19	Clinical characteristics and outcomes in childhood-onset hypertrophic cardiomyopathy. European Heart Journal, 2021, 42, 1988-1996.	1.0	69
20	Computational prediction of protein subdomain stability in MYBPC3 enables clinical risk stratification in hypertrophic cardiomyopathy and enhances variant interpretation. Genetics in Medicine, 2021, 23, 1281-1287.	1.1	11
21	Titin Circular RNAs Create a Back-Splice Motif Essential for SRSF10 Splicing. Circulation, 2021, 143, 1502-1512.	1.6	18
22	Harmonizing the Collection of Clinical Data on Genetic Testing Requisition Forms to Enhance Variant Interpretation in Hypertrophic Cardiomyopathy (HCM). Journal of Molecular Diagnostics, 2021, 23, 589-598.	1.2	5
23	Evidence-Based Assessment of Genes in Dilated Cardiomyopathy. Circulation, 2021, 144, 7-19.	1.6	213
24	Clinical impact of re-evaluating genes and variants implicated in dilated cardiomyopathy. Genetics in Medicine, 2021, 23, 2186-2193.	1.1	17
25	Understanding the genetics of adult-onset dilated cardiomyopathy: what a clinician needs to know. European Heart Journal, 2021, 42, 2384-2396.	1.0	28
26	Prognostic Significance of Nonischemic Myocardial Fibrosis in Patients WithÂNormal LV Volumes and Ejection-Fraction. JACC: Cardiovascular Imaging, 2021, 14, 2353-2365.	2.3	10
27	New Variant With a Previously Unrecognized Mechanism of Pathogenicity in Hypertrophic Cardiomyopathy. Circulation, 2021, 144, 754-757.	1.6	4
28	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2021, 597, E3-E4.	13.7	45
29	Phenotypic Expression and Outcomes in Individuals With Rare Genetic Variants of Hypertrophic Cardiomyopathy. Journal of the American College of Cardiology, 2021, 78, 1097-1110.	1.2	55
30	Worldwide differences in primary prevention implantable cardioverter defibrillator utilization and outcomes in hypertrophic cardiomyopathy. European Heart Journal, 2021, 42, 3932-3944.	1.0	43
31	Shared genetic pathways contribute to risk of hypertrophic and dilated cardiomyopathies with opposite directions of effect. Nature Genetics, 2021, 53, 128-134.	9.4	155
32	Systematic large-scale assessment of the genetic architecture of left ventricular noncompaction reveals diverse etiologies. Genetics in Medicine, 2021, 23, 856-864.	1.1	45
33	The yield of postmortem genetic testing in sudden death cases with structural findings at autopsy. European Journal of Human Genetics, 2020, 28, 17-22.	1.4	38
34	The Egyptian Collaborative Cardiac Genomics (ECCO-GEN) Project: defining a healthy volunteer cohort. Npj Genomic Medicine, 2020, 5, 46.	1.7	5
35	Founder Mutation in N Terminus of Cardiac Troponin I Causes Malignant Hypertrophic Cardiomyopathy. Circulation Genomic and Precision Medicine, 2020, 13, 444-452.	1.6	7
36	Temporal Trend of Age at Diagnosis in Hypertrophic Cardiomyopathy. Circulation: Heart Failure, 2020, 13, e007230.	1.6	48

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37	Spatial and Functional Distribution of MYBPC3 Pathogenic Variants and Clinical Outcomes in Patients With Hypertrophic Cardiomyopathy. Circulation Genomic and Precision Medicine, 2020, 13, 396-405.	1.6	47
38	Genetic and functional insights into the fractal structure of the heart. Nature, 2020, 584, 589-594.	13.7	86
39	Genetic Studies of Hypertrophic Cardiomyopathy in Singaporeans Identify Variants in TNNI3 and TNNT2 That Are Common in Chinese Patients. Circulation Genomic and Precision Medicine, 2020, 13, 424-434.	1.6	18
40	What Is the Risk of Sudden Cardiac Arrest in Inherited Cardiac Conditions?. Journal of the American College of Cardiology, 2020, 75, 2708-2710.	1.2	1
41	The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2020, 581, 434-443.	13.7	6,140
42	Characterising the loss-of-function impact of 5â€™ untranslated region variants in 15,708 individuals. Nature Communications, 2020, 11, 2523.	5.8	99
43	The effect of LRRK2 loss-of-function variants in humans. Nature Medicine, 2020, 26, 869-877.	15.2	79
44	An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. Human Mutation, 2020, 41, 1577-1587.	1.1	10
45	Gene family information facilitates variant interpretation and identification of disease-associated genes in neurodevelopmental disorders. Genome Medicine, 2020, 12, 28.	3.6	42
46	Clinical Features and Natural History of PRKAG2 Variant Cardiac Glycogenosis. Journal of the American College of Cardiology, 2020, 76, 186-197.	1.2	45
47	Predictors of left ventricular remodelling in patients with dilated cardiomyopathy â€“ a cardiovascular magnetic resonance study. European Journal of Heart Failure, 2020, 22, 1160-1170.	2.9	27
48	Reevaluating the Genetic Contribution of Monogenic Dilated Cardiomyopathy. Circulation, 2020, 141, 387-398.	1.6	148
49	An International, Multicentered, Evidence-Based Reappraisal of Genes Reported to Cause Congenital Long QT Syndrome. Circulation, 2020, 141, 418-428.	1.6	238
50	Hypertrophic Cardiomyopathy With Left Ventricular Systolic Dysfunction. Circulation, 2020, 141, 1371-1383.	1.6	108
51	Diagnostic yield of hypertrophic cardiomyopathy in first-degree relatives of decedents with idiopathic left ventricular hypertrophy. Europace, 2020, 22, 632-642.	0.7	20
52	Role of Targeted Therapy in Dilated Cardiomyopathy: The Challenging Road Toward a Personalized Approach. Journal of the American Heart Association, 2019, 8, e012514.	1.6	39
53	Next-generation sequencing using microfluidic PCR enrichment for molecular autopsy. BMC Cardiovascular Disorders, 2019, 19, 174.	0.7	7
54	Association of Titin-Truncating Genetic Variants With Life-threatening Cardiac Arrhythmias in Patients With Dilated Cardiomyopathy and Implanted Defibrillators. JAMA Network Open, 2019, 2, e196520.	2.8	33

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55	Improving the Understanding of Genetic Variants in Rare Disease With Large-scale Reference Populations. JAMA - Journal of the American Medical Association, 2019, 322, 1305.	3.8	7
56	Evaluating the Clinical Validity of Hypertrophic Cardiomyopathy Genes. Circulation Genomic and Precision Medicine, 2019, 12, e002460.	1.6	267
57	Quantitative approaches to variant classification increase the yield and precision of genetic testing in Mendelian diseases: the case of hypertrophic cardiomyopathy. Genome Medicine, 2019, 11, 5.	3.6	90
58	Sudden Death and Left Ventricular Involvement in Arrhythmogenic Cardiomyopathy. Circulation, 2019, 139, 1786-1797.	1.6	122
59	Genetic Variants Associated With Cancer Therapyâ€“Induced Cardiomyopathy. Circulation, 2019, 140, 31-41.	1.6	195
60	Response by Ho et al to Letter Regarding Article, â€œGenotype and Lifetime Burden of Disease in Hypertrophic Cardiomyopathy: Insights From the Sarcomeric Human Cardiomyopathy Registry (SHaRe)â€. Circulation, 2019, 139, 1559-1560.	1.6	4
61	121â€…Re-evaluating the genetic contribution of monogenic dilated cardiomyopathy. , 2019, , .		1
62	Using High-Resolution Variant Frequencies Empowers Clinical Genome Interpretation and Enables Investigation of Genetic Architecture. American Journal of Human Genetics, 2019, 104, 187-190.	2.6	15
63	Withdrawal of pharmacological treatment for heart failure in patients with recovered dilated cardiomyopathy (TRED-HF): an open-label, pilot, randomised trial. Lancet, The, 2019, 393, 61-73.	6.3	379
64	Analysis of 51 proposed hypertrophic cardiomyopathy genes from genome sequencing data in sarcomere negative cases has negligible diagnostic yield. Genetics in Medicine, 2019, 21, 1576-1584.	1.1	44
65	Genomic variant sharing: a position statement. Wellcome Open Research, 2019, 4, 22.	0.9	31
66	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	2.8	93
67	CardioClassifier: disease- and gene-specific computational decision support for clinical genome interpretation. Genetics in Medicine, 2018, 20, 1246-1254.	1.1	75
68	Adaptation and validation of the ACMG/AMP variant classification framework for MYH7-associated inherited cardiomyopathies: recommendations by ClinGenâ€™s Inherited Cardiomyopathy Expert Panel. Genetics in Medicine, 2018, 20, 351-359.	1.1	283
69	Role of titin in cardiomyopathy: from DNA variants to patient stratification. Nature Reviews Cardiology, 2018, 15, 241-252.	6.1	115
70	5â€…Defining the effects of genetic variation using machine learning analysis of CMRS: a study in hypertrophic cardiomyopathy and in a healthy population. , 2018, , .		0
71	Dilated Cardiomyopathy DueÂtoÂBLC2-Associated AthanogeneÂ3Â(BAG3)ÂMutations. Journal of the American College of Cardiology, 2018, 72, 2471-2481.	1.2	93
72	Genotype and Lifetime Burden of Disease in Hypertrophic Cardiomyopathy. Circulation, 2018, 138, 1387-1398.	1.6	468

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73	Genetic Etiology for Alcohol-Induced Cardiac Toxicity. Journal of the American College of Cardiology, 2018, 71, 2293-2302.	1.2	182
74	Reappraisal of Reported Genes for Sudden Arrhythmic Death. Circulation, 2018, 138, 1195-1205.	1.6	271
75	Identification of an INa-dependent and Ito-mediated proarrhythmic mechanism in cardiomyocytes derived from pluripotent stem cells of a Brugada syndrome patient. Scientific Reports, 2018, 8, 11246.	1.6	31
76	Defining the genetic architecture of hypertrophic cardiomyopathy: re-evaluating the role of non-sarcomeric genes. European Heart Journal, 2017, 38, ehw603.	1.0	142
77	Histiocytoid cardiomyopathy and microphthalmia with linear skin defects syndrome: phenotypes linked by truncating variants in NDUFB11. Journal of Physical Education and Sports Management, 2017, 3, a001271.	0.5	19
78	Utility of Post-Mortem Genetic Testing in Cases of Sudden Arrhythmic Death Syndrome. Journal of the American College of Cardiology, 2017, 69, 2134-2145.	1.2	219
79	Truncating Variants in Titin Independently Predict Early Arrhythmias in Patients With Dilated Cardiomyopathy. Journal of the American College of Cardiology, 2017, 69, 2466-2468.	1.2	56
80	Using high-resolution variant frequencies to empower clinical genome interpretation. Genetics in Medicine, 2017, 19, 1151-1158.	1.1	355
81	Phenotype and Clinical Outcomes of TitinÂCardiomyopathy. Journal of the American College of Cardiology, 2017, 70, 2264-2274.	1.2	86
82	Ajmaline blocks I Na and I Kr without eliciting differences between Brugada syndrome patient and control human pluripotent stem cell-derived cardiac clusters. Stem Cell Research, 2017, 25, 233-244.	0.3	25
83	Reassessment of Mendelian gene pathogenicity using 7,855 cardiomyopathy cases and 60,706 reference samples. Genetics in Medicine, 2017, 19, 192-203.	1.1	585
84	Titin-truncating variants affect heart function in disease cohorts and the general population. Nature Genetics, 2017, 49, 46-53.	9.4	255
85	127â€…Relationship between plasma concentrations of b-type natriuretic peptide and exercise capacity in hypertrophic cardiomyopathy. Heart, 2017, 103, A96-A97.	1.2	0
86	50â€…Incremental diagnostic value of cardiovascular magnetic resonance in young adult survivors of sudden cardiac arrest. Heart, 2017, 103, A39-A39.	1.2	1
87	Effects of myosin variants on interacting-heads motif explain distinct hypertrophic and dilated cardiomyopathy phenotypes. ELife, 2017, 6, .	2.8	153
88	Natural genetic variation of the cardiac transcriptome in non-diseased donors and patients with dilated cardiomyopathy. Genome Biology, 2017, 18, 170.	3.8	70
89	Phenotypic and pharmacogenetic evaluation of patients with thiazide-induced hyponatremia. Journal of Clinical Investigation, 2017, 127, 3367-3374.	3.9	58
90	ClinVar data parsing. Wellcome Open Research, 2017, 2, 33.	0.9	19

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91	Recovery of Cardiac Function in Cardiomyopathy Caused by Titin Truncation. JAMA Cardiology, 2016, 1, 234.	3.0	30
92	Analysis of protein-coding genetic variation in 60,706 humans. Nature, 2016, 536, 285-291.	13.7	9,051
93	Loss of RNA expression and allele-specific expression associated with congenital heart disease. Nature Communications, 2016, 7, 12824.	5.8	51
94	Moderate Physical Activity in Healthy Adults Is Associated With Cardiac Remodeling. Circulation: Cardiovascular Imaging, 2016, 9, .	1.3	40
95	Titin truncating mutations: A rare cause of dilated cardiomyopathy in the young. Progress in Pediatric Cardiology, 2016, 40, 41-45.	0.2	23
96	Development of a Comprehensive Sequencing Assay for Inherited Cardiac Condition Genes. Journal of Cardiovascular Translational Research, 2016, 9, 3-11.	1.1	80
97	Shared Genetic Predisposition in Peripartum and Dilated Cardiomyopathies. New England Journal of Medicine, 2016, 374, 233-241.	13.9	432
98	Interpreting de novo Variation in Human Disease Using denovolyzeR. Current Protocols in Human Genetics, 2015, 87, 7.25.1-7.25.15.	3.5	84
99	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. Science, 2015, 350, 1262-1266.	6.0	646
100	Bayesian models for syndrome- and gene-specific probabilities of novel variant pathogenicity. Genome Medicine, 2015, 7, 5.	3.6	22
101	ZBTB17 (MIZ1) Is Important for the Cardiac Stress Response and a Novel Candidate Gene for Cardiomyopathy and Heart Failure. Circulation: Cardiovascular Genetics, 2015, 8, 643-652.	5.1	12
102	Integrated allelic, transcriptional, and phenomic dissection of the cardiac effects of titin truncations in health and disease. Science Translational Medicine, 2015, 7, 270ra6.	5.8	375
103	Detection of mutations in KLHL3 and CUL3 in families with FHHt (familial hyperkalaemic) Tj ETQq1 1 0.784314 rgBT /Ov	1.8	49
104	Paralogue annotation identifies novel pathogenic variants in patients with Brugada syndrome and catecholaminergic polymorphic ventricular tachycardia. Journal of Medical Genetics, 2014, 51, 35-44.	1.5	44
105	NECTAR: a database of codon-centric missense variant annotations. Nucleic Acids Research, 2014, 42, D1013-D1019.	6.5	3
106	Next Generation Diagnostics in Inherited Arrhythmia Syndromes. Journal of Cardiovascular Translational Research, 2013, 6, 94-103.	1.1	31
107	Integrative genomics in cardiovascular medicine. Cardiovascular Research, 2013, 97, 623-630.	1.8	9
108	Towards Clinical Molecular Diagnosis of Inherited Cardiac Conditions: A Comparison of Bench-Top Genome DNA Sequencers. PLoS ONE, 2013, 8, e67744.	1.1	51

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109	Republished review: Next generation sequencing for clinical diagnostics and personalised medicine: implications for the next generation cardiologist. Postgraduate Medical Journal, 2012, 88, 234-239.	0.9	2
110	Next generation sequencing for clinical diagnostics and personalised medicine: implications for the next generation cardiologist. Heart, 2012, 98, 276-281.	1.2	46
111	Paralogous annotation of disease-causing variants in long QT syndrome genes. Human Mutation, 2012, 33, 1188-1191.	1.1	44
112	Endonuclease G is a novel determinant of cardiac hypertrophy and mitochondrial function. Nature, 2011, 478, 114-118.	13.7	135
113	Quality assurance of item writing: During the introduction of multiple choice questions in medicine for high stakes examinations. Medical Teacher, 2009, 31, 238-243.	1.0	38
114	Genomic variant sharing: a position statement. Wellcome Open Research, 0, 4, 22.	0.9	7
115	Effect of taurine administration on symptoms, severity, or clinical outcome of dilated cardiomyopathy and heart failure in humans: a systematic review. Wellcome Open Research, 0, 7, 9.	0.9	1
116	Effect of taurine administration on symptoms, severity, or clinical outcome of dilated cardiomyopathy and heart failure in humans: a systematic review. Wellcome Open Research, 0, 7, 9.	0.9	1
117	Effect of taurine administration on symptoms, severity, or clinical outcome of dilated cardiomyopathy and heart failure in humans: a systematic review. Wellcome Open Research, 0, 7, 9.	0.9	2