James S Ware

List of Publications by Year in Descending Order

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	Exposure to Elevated Nitrogen Dioxide Concentrations and Cardiac Remodeling in Patients With Dilated Cardiomyopathy.. *Journal of Cardiac Failure*, **2022**,	3.3	1
118	Genetic and environmental determinants of diastolic heart function. **2022**, 1, 361-371		0
117	The Gene Curation Coalition: A global effort to harmonize gene-disease evidence resources.. *Genetics in Medicine*, **2022**,	8.1	4
116	Precision Phenotyping of Dilated Cardiomyopathy Using Multidimensional Data. *Journal of the American College of Cardiology*, **2022**, 79, 2219-2232	15.1	2
115	Associations Between Female Sex, Sarcomere Variants, and Clinical Outcomes in Hypertrophic Cardiomyopathy. *Circulation Genomic and Precision Medicine*, **2021**, 14, e003062	5.2	10
114	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.. *Genetics in Medicine*, **2021**,	8.1	0
113	Clinical characteristics and outcomes in childhood-onset hypertrophic cardiomyopathy. *European Heart Journal*, **2021**, 42, 1988-1996	9.5	20
112	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	8.1	3
111	Titin Circular RNAs Create a Back-Splice Motif Essential for SRSF10 Splicing. *Circulation*, **2021**, 143, 1502-1512	16.7	8
110	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. *Journal of Molecular Diagnostics*, **2021**, 23, 589-598	5.1	1
109	Evidence-Based Assessment of Genes in Dilated Cardiomyopathy. *Circulation*, **2021**, 144, 7-19	16.7	34
108	Clinical impact of re-evaluating genes and variants implicated in dilated cardiomyopathy. *Genetics in Medicine*, **2021**, 23, 2186-2193	8.1	3
107	Understanding the genetics of adult-onset dilated cardiomyopathy: what a clinician needs to know. *European Heart Journal*, **2021**, 42, 2384-2396	9.5	6
106	Annotating high-impact 5'untranslated region variants with the UTRannotator. *Bioinformatics*, **2021**, 37, 1171-1173	7.2	8
105	Disease-specific variant pathogenicity prediction significantly improves variant interpretation in inherited cardiac conditions. *Genetics in Medicine*, **2021**, 23, 69-79	8.1	4
104	Enhancing rare variant interpretation in inherited arrhythmias through quantitative analysis of consortium disease cohorts and population controls. *Genetics in Medicine*, **2021**, 23, 47-58	8.1	13
103	Common genetic variants and modifiable risk factors underpin hypertrophic cardiomyopathy susceptibility and expressivity. *Nature Genetics*, **2021**, 53, 135-142	36.3	33

102	Prognostic Significance of Nonischemic Myocardial Fibrosis in Patients With Normal LV Volumes and Ejection-Fraction. *JACC: Cardiovascular Imaging*, **2021**, 14, 2353-2365	8.4	0
101	New Variant With a Previously Unrecognized Mechanism of Pathogenicity in Hypertrophic Cardiomyopathy. *Circulation*, **2021**, 144, 754-757	16.7	1
100	Moderate excess alcohol consumption and adverse cardiac remodelling in dilated cardiomyopathy. *Heart*, **2021**,	5.1	2
99	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. *Nature*, **2021**, 597, E3-E4	50.4	3
98	Phenotypic Expression and Outcomes in Individuals With Rare Genetic Variants of Hypertrophic Cardiomyopathy. *Journal of the American College of Cardiology*, **2021**, 78, 1097-1110	15.1	3
97	Evaluation of gene validity for CPVT and short QT syndrome in sudden arrhythmic death. *European Heart Journal*, **2021**,	9.5	5
96	Worldwide differences in primary prevention implantable cardioverter defibrillator utilization and outcomes in hypertrophic cardiomyopathy. *European Heart Journal*, **2021**, 42, 3932-3944	9.5	6
95	Shared genetic pathways contribute to risk of hypertrophic and dilated cardiomyopathies with opposite directions of effect. *Nature Genetics*, **2021**, 53, 128-134	36.3	35
94	Systematic large-scale assessment of the genetic architecture of left ventricular noncompaction reveals diverse etiologies. *Genetics in Medicine*, **2021**, 23, 856-864	8.1	12
93	The mutational constraint spectrum quantified from variation in 141,456 humans. *Nature*, **2020**, 581, 434-443	50.4	2278
92	Characterising the loss-of-function impact of 5' untranslated region variants in 15,708 individuals. *Nature Communications*, **2020**, 11, 2523	17.4	35
91	The effect of LRRK2 loss-of-function variants in humans. *Nature Medicine*, **2020**, 26, 869-877	50.5	47
90	An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. *Human Mutation*, **2020**, 41, 1577-1587	4.7	4
89	Gene family information facilitates variant interpretation and identification of disease-associated genes in neurodevelopmental disorders. *Genome Medicine*, **2020**, 12, 28	14.4	13
88	Clinical Features and Natural History of PRKAG2 Variant Cardiac Glycogenosis. *Journal of the American College of Cardiology*, **2020**, 76, 186-197	15.1	16
87	Predictors of left ventricular remodelling in patients with dilated cardiomyopathy - a cardiovascular magnetic resonance study. *European Journal of Heart Failure*, **2020**, 22, 1160-1170	12.3	13
86	Reevaluating the Genetic Contribution of Monogenic Dilated Cardiomyopathy. *Circulation*, **2020**, 141, 387-398	16.7	71
85	An International, Multicentered, Evidence-Based Reappraisal of Genes Reported to Cause Congenital Long QT Syndrome. *Circulation*, **2020**, 141, 418-428	16.7	95

84	Diagnostic yield of hypertrophic cardiomyopathy in first-degree relatives of decedents with idiopathic left ventricular hypertrophy. *Europace*, **2020**, 22, 632-642	3.9	10
83	The Egyptian Collaborative Cardiac Genomics (ECCO-GEN) Project: defining a healthy volunteer cohort. *Npj Genomic Medicine*, **2020**, 5, 46	6.2	2
82	Founder Mutation in N Terminus of Cardiac Troponin I Causes Malignant Hypertrophic Cardiomyopathy. *Circulation Genomic and Precision Medicine*, **2020**, 13, 444-452	5.2	3
81	Temporal Trend of Age at Diagnosis in Hypertrophic Cardiomyopathy: An Analysis of the International Sarcomeric Human Cardiomyopathy Registry. *Circulation: Heart Failure*, **2020**, 13, e007230	7.6	16
80	Spatial and Functional Distribution of Pathogenic Variants and Clinical Outcomes in Patients With Hypertrophic Cardiomyopathy. *Circulation Genomic and Precision Medicine*, **2020**, 13, 396-405	5.2	19
79	Genetic and functional insights into the fractal structure of the heart. *Nature*, **2020**, 584, 589-594	50.4	26
78	Genetic Studies of Hypertrophic Cardiomyopathy in Singaporeans Identify Variants in and That Are Common in Chinese Patients. *Circulation Genomic and Precision Medicine*, **2020**, 13, 424-434	5.2	4
77	The yield of postmortem genetic testing in sudden death cases with structural findings at autopsy. *European Journal of Human Genetics*, **2020**, 28, 17-22	5.3	16
76	Hypertrophic Cardiomyopathy With Left Ventricular Systolic Dysfunction: Insights From the SHaRe Registry. *Circulation*, **2020**, 141, 1371-1383	16.7	43
75	Improving the Understanding of Genetic Variants in Rare Disease With Large-scale Reference Populations. *JAMA - Journal of the American Medical Association*, **2019**, 322, 1305-1306	27.4	3
74	Evaluating the Clinical Validity of Hypertrophic Cardiomyopathy Genes. *Circulation Genomic and Precision Medicine*, **2019**, 12, e002460	5.2	132
73	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	14.4	54
72	Sudden Death and Left Ventricular Involvement in Arrhythmogenic Cardiomyopathy. *Circulation*, **2019**, 139, 1786-1797	16.7	70
71	Genetic Variants Associated With Cancer Therapy-Induced Cardiomyopathy. *Circulation*, **2019**, 140, 31-41	16.7	110
70	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)". *Circulation*, **2019**, 139, 1559-1560	16.7	2
69	Role of Targeted Therapy in Dilated Cardiomyopathy: The Challenging Road Toward a Personalized Approach. *Journal of the American Heart Association*, **2019**, 8, e012514	6	21
68	Next-generation sequencing using microfluidic PCR enrichment for molecular autopsy. *BMC Cardiovascular Disorders*, **2019**, 19, 174	2.3	5
67	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	19.4	20

66	Genomic variant sharing: a position statement. *Wellcome Open Research*, **2019**, 4, 22	4.8	16
65	Using High-Resolution Variant Frequencies Empowers Clinical Genome Interpretation and Enables Investigation of Genetic Architecture. *American Journal of Human Genetics*, **2019**, 104, 187-190	11	8
64	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	40	198
63	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	8.1	25
62	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. *Annals of Neurology*, **2018**, 83, 1105-1124	9.4	59
61	CardioClassifier: disease- and gene-specific computational decision support for clinical genome interpretation. *Genetics in Medicine*, **2018**, 20, 1246-1254	8.1	45
60	Adaptation and validation of the ACMG/AMP variant classification framework for MYH7-associated inherited cardiomyopathies: recommendations by ClinGen's Inherited Cardiomyopathy Expert Panel. *Genetics in Medicine*, **2018**, 20, 351-359	8.1	173
59	Reappraisal of Reported Genes for Sudden Arrhythmic Death: Evidence-Based Evaluation of Gene Validity for Brugada Syndrome. *Circulation*, **2018**, 138, 1195-1205	16.7	158
58	Identification of an I-dependent and I-mediated proarrhythmic mechanism in cardiomyocytes derived from pluripotent stem cells of a Brugada syndrome patient. *Scientific Reports*, **2018**, 8, 11246	4.9	20
57	Role of titin in cardiomyopathy: from DNA variants to patient stratification. *Nature Reviews Cardiology*, **2018**, 15, 241-252	14.8	69
56	Dilated Cardiomyopathy Due to BLC2-Associated Athanogene 3 (BAG3) Mutations. *Journal of the American College of Cardiology*, **2018**, 72, 2471-2481	15.1	53
55	Genotype and Lifetime Burden of Disease in Hypertrophic Cardiomyopathy: Insights from the Sarcomeric Human Cardiomyopathy Registry (SHaRe). *Circulation*, **2018**, 138, 1387-1398	16.7	210
54	Genetic Etiology for Alcohol-Induced Cardiac Toxicity. *Journal of the American College of Cardiology*, **2018**, 71, 2293-2302	15.1	112
53	Defining the genetic architecture of hypertrophic cardiomyopathy: re-evaluating the role of non-sarcomeric genes. *European Heart Journal*, **2017**, 38, 3461-3468	9.5	101
52	Histiocytoid cardiomyopathy and microphthalmia with linear skin defects syndrome: phenotypes linked by truncating variants in. *Journal of Physical Education and Sports Management*, **2017**, 3, a001271	2.8	15
51	Utility of Post-Mortem Genetic Testing in Cases of Sudden Arrhythmic Death Syndrome. *Journal of the American College of Cardiology*, **2017**, 69, 2134-2145	15.1	126
50	Truncating Variants in Titin Independently Predict Early Arrhythmias in Patients With Dilated Cardiomyopathy. *Journal of the American College of Cardiology*, **2017**, 69, 2466-2468	15.1	40
49	Using high-resolution variant frequencies to empower clinical genome interpretation. *Genetics in Medicine*, **2017**, 19, 1151-1158	8.1	208

48	Phenotype and Clinical Outcomes of Titin Cardiomyopathy. *Journal of the American College of Cardiology*, **2017**, 70, 2264-2274	15.1	57
47	Natural genetic variation of the cardiac transcriptome in non-diseased donors and patients with dilated cardiomyopathy. *Genome Biology*, **2017**, 18, 170	18.3	40
46	Ajmaline blocks I and I without eliciting differences between Brugada syndrome patient and control human pluripotent stem cell-derived cardiac clusters. *Stem Cell Research*, **2017**, 25, 233-244	1.6	17
45	Reassessment of Mendelian gene pathogenicity using 7,855 cardiomyopathy cases and 60,706 reference samples. *Genetics in Medicine*, **2017**, 19, 192-203	8.1	386
44	Titin-truncating variants affect heart function in disease cohorts and the general population. *Nature Genetics*, **2017**, 49, 46-53	36.3	179
43	127 Relationship between plasma concentrations of b-type natriuretic peptide and exercise capacity in hypertrophic cardiomyopathy. *Heart*, **2017**, 103, A96-A97	5.1	
42	50 Incremental diagnostic value of cardiovascular magnetic resonance in young adult survivors of sudden cardiac arrest. *Heart*, **2017**, 103, A39-A39	5.1	1
41	Effects of myosin variants on interacting-heads motif explain distinct hypertrophic and dilated cardiomyopathy phenotypes. *ELife*, **2017**, 6,	8.9	87
40	Phenotypic and pharmacogenetic evaluation of patients with thiazide-induced hyponatremia. *Journal of Clinical Investigation*, **2017**, 127, 3367-3374	15.9	35
39	ClinVar data parsing. *Wellcome Open Research*, **2017**, 2, 33	4.8	16
38	Analysis of protein-coding genetic variation in 60,706 humans. *Nature*, **2016**, 536, 285-91	50.4	6940
37	Loss of RNA expression and allele-specific expression associated with congenital heart disease. *Nature Communications*, **2016**, 7, 12824	17.4	33
36	Moderate Physical Activity in Healthy Adults Is Associated With Cardiac Remodeling. *Circulation: Cardiovascular Imaging*, **2016**, 9,	3.9	27
35	Titin truncating mutations: A rare cause of dilated cardiomyopathy in the young. *Progress in Pediatric Cardiology*, **2016**, 40, 41-45	0.4	17
34	Development of a Comprehensive Sequencing Assay for Inherited Cardiac Condition Genes. *Journal of Cardiovascular Translational Research*, **2016**, 9, 3-11	3.3	63
33	Shared Genetic Predisposition in Peripartum and Dilated Cardiomyopathies. *New England Journal of Medicine*, **2016**, 374, 233-41	59.2	290
32	Recovery of Cardiac Function in Cardiomyopathy Caused by Titin Truncation. *JAMA Cardiology*, **2016**, 1, 234-5	16.2	21
31	Bayesian models for syndrome- and gene-specific probabilities of novel variant pathogenicity. *Genome Medicine*, **2015**, 7, 5	14.4	19

30	ZBTB17 (MIZ1) Is Important for the Cardiac Stress Response and a Novel Candidate Gene for Cardiomyopathy and Heart Failure. *Circulation: Cardiovascular Genetics*, **2015**, 8, 643-52		9
29	Integrated allelic, transcriptional, and phenomic dissection of the cardiac effects of titin truncations in health and disease. *Science Translational Medicine*, **2015**, 7, 270ra6	17.5	267
28	Interpreting de novo Variation in Human Disease Using denovolyzeR. *Current Protocols in Human Genetics*, **2015**, 87, 7.25.1-7.25.15	3.2	56
27	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. *Science*, **2015**, 350, 1262-6	33.3	406
26	Detection of mutations in KLHL3 and CUL3 in families with FHHt (familial hyperkalaemic hypertension or Gordon's syndrome). *Clinical Science*, **2014**, 126, 721-6	6.5	38
25	Paralogue annotation identifies novel pathogenic variants in patients with Brugada syndrome and catecholaminergic polymorphic ventricular tachycardia. *Journal of Medical Genetics*, **2014**, 51, 35-44	5.8	31
24	NECTAR: a database of codon-centric missense variant annotations. *Nucleic Acids Research*, **2014**, 42, D1013-9	20.1	2
23	Next generation diagnostics in inherited arrhythmia syndromes : a comparison of two approaches. *Journal of Cardiovascular Translational Research*, **2013**, 6, 94-103	3.3	28
22	Integrative genomics in cardiovascular medicine. *Cardiovascular Research*, **2013**, 97, 623-30	9.9	8
21	Towards clinical molecular diagnosis of inherited cardiac conditions: a comparison of bench-top genome DNA sequencers. *PLoS ONE*, **2013**, 8, e67744	3.7	48
20	Republished review: Next generation sequencing for clinical diagnostics and personalised medicine: implications for the next generation cardiologist. *Postgraduate Medical Journal*, **2012**, 88, 234-9	2	2
19	Next generation sequencing for clinical diagnostics and personalised medicine: implications for the next generation cardiologist. *Heart*, **2012**, 98, 276-81	5.1	39
18	Paralogous annotation of disease-causing variants in long QT syndrome genes. *Human Mutation*, **2012**, 33, 1188-1191	4.7	26
17	Endonuclease G is a novel determinant of cardiac hypertrophy and mitochondrial function. *Nature*, **2011**, 478, 114-8	50.4	114
16	Quality assurance of item writing: during the introduction of multiple choice questions in medicine for high stakes examinations. *Medical Teacher*, **2009**, 31, 238-43	3	31
15	Effect of taurine administration on symptoms, severity, or clinical outcome of dilated cardiomyopathy and heart failure in humans: a systematic review. *Wellcome Open Research*,7, 9	4.8	
14	Effect of taurine administration on symptoms, severity, or clinical outcome of dilated cardiomyopathy and heart failure in humans: a systematic review. *Wellcome Open Research*,7, 9	4.8	
13	Genomic variant sharing: a position statement. *Wellcome Open Research*,4, 22	4.8	5

12	Quantitative approaches to variant classification increase the yield and precision of genetic testing in Mendelian diseases: The case of hypertrophic cardiomyopathy	1
11	Genomics of Egyptian Healthy Volunteers: The EHVol Study	1
10	Reassessment of Mendelian gene pathogenicity using 7,855 cardiomyopathy cases and 60,706 reference samples	5
9	Using high-resolution variant frequencies to empower clinical genome interpretation	4
8	Gene family information facilitates variant interpretation and identification of disease-associated genes	6
7	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
6	Analysis of HCM in an understudied population reveals a new mechanism of pathogenicity	2
5	The mutational constraint spectrum quantified from variation in 141,456 humans	381
4	Characterising the loss-of-function impact of 5′ untranslated region variants in whole genome sequence data from 15,708 individuals	5
3	Human loss-of-function variants suggest that partial LRRK2 inhibition is a safe therapeutic strategy for Parkinson's disease	7
2	Genetic and environmental determinants of diastolic heart function	1
1	Outcomes and phenotypic expression of rare variants in hypertrophic cardiomyopathy genes amongst UK Biobank participants	1