# James S Ware

List of Publications by Year
in descending order



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|---|---|---|---|
| 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 | Analysis of protein-coding genetic variation in 60,706 humans. Nature, 2016, 536, 285-291. | 13.7 | 9,051 |
| 2 | The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2020, 581, 434-443. | 13.7 | 6,140 |
| 3 | De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. Science, 2015, 350, 1262-1266. | 6.0 | 646 |
| 4 | 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 |
| 5 | Genotype and Lifetime Burden of Disease in Hypertrophic Cardiomyopathy. Circulation, 2018, 138, 1387-1398. | 1.6 | 468 |
| 6 | Shared Genetic Predisposition in Peripartum and Dilated Cardiomyopathies. New England Journal of Medicine, 2016, 374, 233-241. | 13.9 | 432 |
| 7 | 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 |
| 8 | 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 |
| 9 | Using high-resolution variant frequencies to empower clinical genome interpretation. Genetics in Medicine, 2017, 19, 1151-1158. | 1.1 | 355 |
| 10 | 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 |
| 11 | Reappraisal of Reported Genes for Sudden Arrhythmic Death. Circulation, 2018, 138, 1195-1205. | 1.6 | 271 |
| 12 | Evaluating the Clinical Validity of Hypertrophic Cardiomyopathy Genes. Circulation Genomic and Precision Medicine, 2019, 12, e002460. | 1.6 | 267 |
| 13 | Titin-truncating variants affect heart function in disease cohorts and the general population. Nature Genetics, 2017, 49, 46-53. | 9.4 | 255 |
| 14 | An International, Multicentered, Evidence-Based Reappraisal of Genes Reported to Cause Congenital Long QT Syndrome. Circulation, 2020, 141, 418-428. | 1.6 | 238 |
| 15 | 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 |
| 16 | Evidence-Based Assessment of Genes in Dilated Cardiomyopathy. Circulation, 2021, 144, 7-19. | 1.6 | 213 |
| 17 | Genetic Variants Associated With Cancer Therapyâ€“Induced Cardiomyopathy. Circulation, 2019, 140, 31-41. | 1.6 | 195 |
| 18 | Genetic Etiology for Alcohol-Induced Cardiac Toxicity. Journal of the American College of Cardiology, 2018, 71, 2293-2302. | 1.2 | 182 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Common genetic variants and modifiable risk factors underpin hypertrophic cardiomyopathy susceptibility and expressivity. Nature Genetics, 2021, 53, 135-142. | 9.4 | 165 |
| 20 | 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 |
| 21 | Effects of myosin variants on interacting-heads motif explain distinct hypertrophic and dilated cardiomyopathy phenotypes. ELife, 2017, 6, . | 2.8 | 153 |
| 22 | Reevaluating the Genetic Contribution of Monogenic Dilated Cardiomyopathy. Circulation, 2020, 141, 387-398. | 1.6 | 148 |
| 23 | Defining the genetic architecture of hypertrophic cardiomyopathy: re-evaluating the role of non-sarcomeric genes. European Heart Journal, 2017, 38, ehw603. | 1.0 | 142 |
| 24 | Endonuclease G is a novel determinant of cardiac hypertrophy and mitochondrial function. Nature, 2011, 478, 114-118. | 13.7 | 135 |
| 25 | Sudden Death and Left Ventricular Involvement in Arrhythmogenic Cardiomyopathy. Circulation, 2019, 139, 1786-1797. | 1.6 | 122 |
| 26 | Role of titin in cardiomyopathy: from DNA variants to patient stratification. Nature Reviews Cardiology, 2018, 15, 241-252. | 6.1 | 115 |
| 27 | Hypertrophic Cardiomyopathy With Left Ventricular Systolic Dysfunction. Circulation, 2020, 141, 1371-1383. | 1.6 | 108 |
| 28 | 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 |
| 29 | Characterising the loss-of-function impact of 5â€™ untranslated region variants in 15,708 individuals. Nature Communications, 2020, 11, 2523. | 5.8 | 99 |
| 30 | Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124. | 2.8 | 93 |
| 31 | Dilated Cardiomyopathy DueÂtoÂBLC2-Associated AthanogeneÂ3Â(BAG3)ÂMutations. Journal of the American College of Cardiology, 2018, 72, 2471-2481. | 1.2 | 93 |
| 32 | 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 |
| 33 | Phenotype and Clinical Outcomes of TitinÂCardiomyopathy. Journal of the American College of Cardiology, 2017, 70, 2264-2274. | 1.2 | 86 |
| 34 | Genetic and functional insights into the fractal structure of the heart. Nature, 2020, 584, 589-594. | 13.7 | 86 |
| 35 | 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 |
| 36 | Development of a Comprehensive Sequencing Assay for Inherited Cardiac Condition Genes. Journal of Cardiovascular Translational Research, 2016, 9, 3-11. | 1.1 | 80 |

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|---|---|---|---|
| 37 | The effect of LRRK2 loss-of-function variants in humans. Nature Medicine, 2020, 26, 869-877. | 15.2 | 79 |
| 38 | 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 |
| 39 | CardioClassifier: disease- and gene-specific computational decision support for clinical genome interpretation. Genetics in Medicine, 2018, 20, 1246-1254. | 1.1 | 75 |
| 40 | Natural genetic variation of the cardiac transcriptome in non-diseased donors and patients with dilated cardiomyopathy. Genome Biology, 2017, 18, 170. | 3.8 | 70 |
| 41 | Clinical characteristics and outcomes in childhood-onset hypertrophic cardiomyopathy. European Heart Journal, 2021, 42, 1988-1996. | 1.0 | 69 |
| 42 | Phenotypic and pharmacogenetic evaluation of patients with thiazide-induced hyponatremia. Journal of Clinical Investigation, 2017, 127, 3367-3374. | 3.9 | 58 |
| 43 | 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 |
| 44 | Evaluation of gene validity for CPVT and short QT syndrome in sudden arrhythmic death. European Heart Journal, 2022, 43, 1500-1510. | 1.0 | 57 |
| 45 | 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 |
| 46 | The Gene Curation Coalition: A global effort to harmonize geneâ€“disease evidence resources. Genetics in Medicine, 2022, 24, 1732-1742. | 1.1 | 56 |
| 47 | 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 |
| 48 | Towards Clinical Molecular Diagnosis of Inherited Cardiac Conditions: A Comparison of Bench-Top Genome DNA Sequencers. PLoS ONE, 2013, 8, e67744. | 1.1 | 51 |
| 49 | Loss of RNA expression and allele-specific expression associated with congenital heart disease. Nature Communications, 2016, 7, 12824. | 5.8 | 51 |
| 50 | Detection of mutations in KLHL3 and CUL3 in families with FHHt (familial hyperkalaemic) Tj ETQq0 0 0 rgBT /Overlock 10 T | 1.8 | 49 |
| 51 | Temporal Trend of Age at Diagnosis in Hypertrophic Cardiomyopathy. Circulation: Heart Failure, 2020, 13, e007230. | 1.6 | 48 |
| 52 | 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 |
| 53 | Next generation sequencing for clinical diagnostics and personalised medicine: implications for the next generation cardiologist. Heart, 2012, 98, 276-281. | 1.2 | 46 |
| 54 | Clinical Features and Natural History of PRKAG2 Variant Cardiac Glycogenosis. Journal of the American College of Cardiology, 2020, 76, 186-197. | 1.2 | 45 |

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|---|---|---|---|
| 55 | Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2021, 597, E3-E4. | 13.7 | 45 |
| 56 | 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 |
| 57 | Paralogous annotation of disease-causing variants in long QT syndrome genes. Human Mutation, 2012, 33, 1188-1191. | 1.1 | 44 |
| 58 | 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 |
| 59 | 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 |
| 60 | Worldwide differences in primary prevention implantable cardioverter defibrillator utilization and outcomes in hypertrophic cardiomyopathy. European Heart Journal, 2021, 42, 3932-3944. | 1.0 | 43 |
| 61 | Gene family information facilitates variant interpretation and identification of disease-associated genes in neurodevelopmental disorders. Genome Medicine, 2020, 12, 28. | 3.6 | 42 |
| 62 | Moderate Physical Activity in Healthy Adults Is Associated With Cardiac Remodeling. Circulation: Cardiovascular Imaging, 2016, 9, . | 1.3 | 40 |
| 63 | 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 |
| 64 | Disease-specific variant pathogenicity prediction significantly improves variant interpretation in inherited cardiac conditions. Genetics in Medicine, 2021, 23, 69-79. | 1.1 | 39 |
| 65 | 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 |
| 66 | 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 |
| 67 | Associations Between Female Sex, Sarcomere Variants, and Clinical Outcomes in Hypertrophic Cardiomyopathy. Circulation Genomic and Precision Medicine, 2021, 14, e003062. | 1.6 | 38 |
| 68 | 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 |
| 69 | Next Generation Diagnostics in Inherited Arrhythmia Syndromes. Journal of Cardiovascular Translational Research, 2013, 6, 94-103. | 1.1 | 31 |
| 70 | 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 |
| 71 | Genomic variant sharing: a position statement. Wellcome Open Research, 2019, 4, 22. | 0.9 | 31 |
| 72 | Recovery of Cardiac Function in Cardiomyopathy Caused by Titin Truncation. JAMA Cardiology, 2016, 1, 234. | 3.0 | 30 |

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|---|---|---|---|
| 73 | Understanding the genetics of adult-onset dilated cardiomyopathy: what a clinician needs to know. European Heart Journal, 2021, 42, 2384-2396. | 1.0 | 28 |
| 74 | 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 |
| 75 | Annotating high-impact 5â€²untranslated region variants with the UTRannotator. Bioinformatics, 2021, 37, 1171-1173. | 1.8 | 27 |
| 76 | 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 |
| 77 | 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 |
| 78 | Precision Phenotyping of Dilated Cardiomyopathy Using Multidimensional Data. Journal of the American College of Cardiology, 2022, 79, 2219-2232. | 1.2 | 24 |
| 79 | Titin truncating mutations: A rare cause of dilated cardiomyopathy in the young. Progress in Pediatric Cardiology, 2016, 40, 41-45. | 0.2 | 23 |
| 80 | Bayesian models for syndrome- and gene-specific probabilities of novel variant pathogenicity. Genome Medicine, 2015, 7, 5. | 3.6 | 22 |
| 81 | Diagnostic yield of hypertrophic cardiomyopathy in first-degree relatives of decedents with idiopathic left ventricular hypertrophy. Europace, 2020, 22, 632-642. | 0.7 | 20 |
| 82 | 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 |
| 83 | ClinVar data parsing. Wellcome Open Research, 2017, 2, 33. | 0.9 | 19 |
| 84 | 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 |
| 85 | Titin Circular RNAs Create a Back-Splice Motif Essential for SRSF10 Splicing. Circulation, 2021, 143, 1502-1512. | 1.6 | 18 |
| 86 | Clinical impact of re-evaluating genes and variants implicated in dilated cardiomyopathy. Genetics in Medicine, 2021, 23, 2186-2193. | 1.1 | 17 |
| 87 | 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 |
| 88 | 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 |
| 89 | 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 |
| 90 | Genetic and environmental determinants of diastolic heart function. , 2022, 1, 361-371. | | 12 |

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|---|---|---|---|
| 91 | 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 |
| 92 | An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. Human Mutation, 2020, 41, 1577-1587. | 1.1 | 10 |
| 93 | Prognostic Significance of Nonischemic Myocardial Fibrosis in Patients WithÂNormal LV Volumes and Ejection-Fraction. JACC: Cardiovascular Imaging, 2021, 14, 2353-2365. | 2.3 | 10 |
| 94 | Integrative genomics in cardiovascular medicine. Cardiovascular Research, 2013, 97, 623-630. | 1.8 | 9 |
| 95 | Next-generation sequencing using microfluidic PCR enrichment for molecular autopsy. BMC Cardiovascular Disorders, 2019, 19, 174. | 0.7 | 7 |
| 96 | 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 |
| 97 | 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 |
| 98 | Genomic variant sharing: a position statement. Wellcome Open Research, 0, 4, 22. | 0.9 | 7 |
| 99 | Moderate excess alcohol consumption and adverse cardiac remodelling in dilated cardiomyopathy. Heart, 2022, 108, 619-625. | 1.2 | 6 |
| 100 | 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 |
| 101 | The Egyptian Collaborative Cardiac Genomics (ECCO-GEN) Project: defining a healthy volunteer cohort. Npj Genomic Medicine, 2020, 5, 46. | 1.7 | 5 |
| 102 | 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 |
| 103 | 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 |
| 104 | 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 |
| 105 | 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 |
| 106 | New Variant With a Previously Unrecognized Mechanism of Pathogenicity in Hypertrophic Cardiomyopathy. Circulation, 2021, 144, 754-757. | 1.6 | 4 |
| 107 | NECTAR: a database of codon-centric missense variant annotations. Nucleic Acids Research, 2014, 42, D1013-D1019. | 6.5 | 3 |
| 108 | Direct and indirect effect of the COVID-19 pandemic on patients with cardiomyopathy. Open Heart, 2022, 9, e001918. | 0.9 | 3 |

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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 | 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 |
| 111 | 50â€…Incremental diagnostic value of cardiovascular magnetic resonance in young adult survivors of sudden cardiac arrest. Heart, 2017, 103, A39-A39. | 1.2 | 1 |
| 112 | 121â€…Re-evaluating the genetic contribution of monogenic dilated cardiomyopathy. , 2019, , . | | 1 |
| 113 | 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 |
| 114 | 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 |
| 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 | 127â€…Relationship between plasma concentrations of b-type natriuretic peptide and exercise capacity in hypertrophic cardiomyopathy. Heart, 2017, 103, A96-A97. | 1.2 | 0 |
| 117 | 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 |