Roddy Walsh

List of Publications by Year
in descending order

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71 papers	4,941 citations	126907 33 h-index	106344 65 g-index
81 all docs	81 docs citations	81 times ranked	6562 citing authors

#	Article	IF	Citations
1	Reassessment of Mendelian gene pathogenicity using 7,855 cardiomyopathy cases and 60,706 reference samples. Genetics in Medicine, 2017, 19, 192-203.	2.4	585
2	Integrated allelic, transcriptional, and phenomic dissection of the cardiac effects of titin truncations in health and disease. Science Translational Medicine, 2015, 7, 270ra6.	12.4	375
3	Using high-resolution variant frequencies to empower clinical genome interpretation. Genetics in Medicine, 2017, 19, 1151-1158.	2.4	355
4	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.	2.4	283
5	Evaluating the Clinical Validity of Hypertrophic Cardiomyopathy Genes. Circulation Genomic and Precision Medicine, 2019, 12, e002460.	3.6	267
6	Titin-truncating variants affect heart function in disease cohorts and the general population. Nature Genetics, 2017, 49, 46-53.	21.4	255
7	Evidence-Based Assessment of Genes in Dilated Cardiomyopathy. Circulation, 2021, 144, 7-19.	1.6	213
8	Genetic Variants Associated With Cancer Therapyâ€“Induced Cardiomyopathy. Circulation, 2019, 140, 31-41.	1.6	195
9	Genetic Etiology for Alcohol-Induced Cardiac Toxicity. Journal of the American College of Cardiology, 2018, 71, 2293-2302.	2.8	182
10	Shared genetic pathways contribute to risk of hypertrophic and dilated cardiomyopathies with opposite directions of effect. Nature Genetics, 2021, 53, 128-134.	21.4	155
11	Reevaluating the Genetic Contribution of Monogenic Dilated Cardiomyopathy. Circulation, 2020, 141, 387-398.	1.6	148
12	Defining the genetic architecture of hypertrophic cardiomyopathy: re-evaluating the role of non-sarcomeric genes. European Heart Journal, 2017, 38, ehw603.	2.2	142
13	Dilated Cardiomyopathy DueÂtoÂBLC2-Associated AthanogeneÂ3Â(BAG3)ÂMutations. Journal of the American College of Cardiology, 2018, 72, 2471-2481.	2.8	93
14	Complex roads from genotype to phenotype in dilated cardiomyopathy: scientific update from the Working Group of Myocardial Function of the European Society of Cardiology. Cardiovascular Research, 2018, 114, 1287-1303.	3.8	91
15	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.	8.2	90
16	Phenotype and Clinical Outcomes of TitinÂCardiomyopathy. Journal of the American College of Cardiology, 2017, 70, 2264-2274.	2.8	86
17	Transethnic Genome-Wide Association Study Provides Insights in the Genetic Architecture and Heritability of Long QT Syndrome. Circulation, 2020, 142, 324-338.	1.6	83
18	Development of a Comprehensive Sequencing Assay for Inherited Cardiac Condition Genes. Journal of Cardiovascular Translational Research, 2016, 9, 3-11.	2.4	80

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19	CardioClassifier: disease- and gene-specific computational decision support for clinical genome interpretation. Genetics in Medicine, 2018, 20, 1246-1254.	2.4	75
20	Enhancing rare variant interpretation in inherited arrhythmias through quantitative analysis of consortium disease cohorts and population controls. Genetics in Medicine, 2021, 23, 47-58.	2.4	57
21	Evaluation of gene validity for CPVT and short QT syndrome in sudden arrhythmic death. European Heart Journal, 2022, 43, 1500-1510.	2.2	57
22	Truncating Variants in Titin Independently Predict Early Arrhythmias in Patients With Dilated Cardiomyopathy. Journal of the American College of Cardiology, 2017, 69, 2466-2468.	2.8	56
23	Phenotypic Expression and Outcomes in Individuals With Rare Genetic Variants of Hypertrophic Cardiomyopathy. Journal of the American College of Cardiology, 2021, 78, 1097-1110.	2.8	55
24	Genome-wide association analyses identify new Brugada syndrome risk loci and highlight a new mechanism of sodium channel regulation in disease susceptibility. Nature Genetics, 2022, 54, 232-239.	21.4	55
25	Defining the diagnostic effectiveness of genes for inclusion in panels: the experience of two decades of genetic testing for hypertrophic cardiomyopathy at a single center. Genetics in Medicine, 2019, 21, 284-292.	2.4	54
26	Towards Clinical Molecular Diagnosis of Inherited Cardiac Conditions: A Comparison of Bench-Top Genome DNA Sequencers. PLoS ONE, 2013, 8, e67744.	2.5	51
27	Minor hypertrophic cardiomyopathy genes, major insights into the genetics of cardiomyopathies. Nature Reviews Cardiology, 2022, 19, 151-167.	13.7	50
28	Detection of mutations in KLHL3 and CUL3 in families with FHHt (familial hyperkalaemic) Tj ETQq0 0 0 rgBT /Overlock 10 T	4.3	49
29	Systematic large-scale assessment of the genetic architecture of left ventricular noncompaction reveals diverse etiologies. Genetics in Medicine, 2021, 23, 856-864.	2.4	45
30	Paralogous annotation of disease-causing variants in long QT syndrome genes. Human Mutation, 2012, 33, 1188-1191.	2.5	44
31	Paralogue annotation identifies novel pathogenic variants in patients with Brugada syndrome and catecholaminergic polymorphic ventricular tachycardia. Journal of Medical Genetics, 2014, 51, 35-44.	3.2	44
32	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.	2.4	44
33	Anti-inflammatory modulation of chronic airway inflammation in the murine house dust mite model. Pulmonary Pharmacology and Therapeutics, 2008, 21, 637-647.	2.6	42
34	Contemporary Insights Into the Genetics of Hypertrophic Cardiomyopathy: Toward a New Era in Clinical Testing?. Journal of the American Heart Association, 2020, 9, e015473.	3.7	42
35	Moderate Physical Activity in Healthy Adults Is Associated With Cardiac Remodeling. Circulation: Cardiovascular Imaging, 2016, 9, .	2.6	40
36	When genetic burden reaches threshold. European Heart Journal, 2020, 41, 3849-3855.	2.2	40

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37	Disease-specific variant pathogenicity prediction significantly improves variant interpretation in inherited cardiac conditions. Genetics in Medicine, 2021, 23, 69-79.	2.4	39
38	Recovery of Cardiac Function in Cardiomyopathy Caused by Titin Truncation. JAMA Cardiology, 2016, 1, 234.	6.1	30
39	Titin truncating mutations: A rare cause of dilated cardiomyopathy in the young. Progress in Pediatric Cardiology, 2016, 40, 41-45.	0.4	23
40	Common and rare susceptibility genetic variants predisposing to Brugada syndrome in Thailand. Heart Rhythm, 2020, 17, 2145-2153.	0.7	23
41	Bayesian models for syndrome- and gene-specific probabilities of novel variant pathogenicity. Genome Medicine, 2015, 7, 5.	8.2	22
42	Targeted therapies in genetic dilated and hypertrophic cardiomyopathies: from molecular mechanisms to therapeutic targets. A position paper from the Heart Failure Association (HFA) and the Working Group on Myocardial Function of the European Society of Cardiology (ESC). European Journal of Heart Failure, 2022, 24, 406-420.	7.1	22
43	Advantages and Perils of Clinical Whole-Exome and Whole-Genome Sequencing in Cardiomyopathy. Cardiovascular Drugs and Therapy, 2020, 34, 241-253.	2.6	21
44	Variant Intronic Enhancer Controls SCN10A-short Expression and Heart Conduction. Circulation, 2021, 144, 229-242.	1.6	20
45	Time Course Toxicogenomic Profiles in CD-1 Mice after Nontoxic and Nonlethal Hepatotoxic Paracetamol Administration. Chemical Research in Toxicology, 2004, 17, 1551-1561.	3.3	19
46	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.	1.2	19
47	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.	3.6	18
48	Using High-Resolution Variant Frequencies Empowers Clinical Genome Interpretation and Enables Investigation of Genetic Architecture. American Journal of Human Genetics, 2019, 104, 187-190.	6.2	15
49	Illuminating the path from genetics to clinical outcome in Brugada syndrome. European Heart Journal, 2021, 42, 1091-1093.	2.2	12
50	An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. Human Mutation, 2020, 41, 1577-1587.	2.5	10
51	Issues and Challenges in Diagnostic Sequencing for Inherited Cardiac Conditions. Clinical Chemistry, 2017, 63, 116-128.	3.2	7
52	Rare variants in KDR, encoding VEGF Receptor 2, are associated with tetralogy of Fallot. Genetics in Medicine, 2021, 23, 1952-1960.	2.4	7
53	Genetic modifiers to the PLN L39X mutation in a patient with DCM and sustained ventricular tachycardia?. Global Cardiology Science & Practice, 2015, 2015, 29.	0.4	6
54	SCN5A variants in Brugada syndrome: True, true false, or false true. Journal of Cardiovascular Electrophysiology, 2019, 30, 128-131.	1.7	5

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55	The Egyptian Collaborative Cardiac Genomics (ECCO-GEN) Project: defining a healthy volunteer cohort. Npj Genomic Medicine, 2020, 5, 46.	3.8	5
56	143â€…Clinical and Genetic Characteristics of Familial Dilated Cardiomyopathy in a Large UK Prospective Cohort: Abstract 143 Table 1. Heart, 2016, 102, A103-A104.	2.9	4
57	ALPK3: a full spectrum cardiomyopathy gene?. European Heart Journal, 2021, 42, 3074-3077.	2.2	4
58	New Variant With a Previously Unrecognized Mechanism of Pathogenicity in Hypertrophic Cardiomyopathy. Circulation, 2021, 144, 754-757.	1.6	4
59	NECTAR: a database of codon-centric missense variant annotations. Nucleic Acids Research, 2014, 42, D1013-D1019.	14.5	3
60	Predicting Risk for Adult-Onset Sudden Cardiac Death in the Population. Journal of the American College of Cardiology, 2019, 74, 2635-2637.	2.8	3
61	Desmin variants in cardiomyopathies â€“ the hard yards in defining pathogenicity. International Journal of Cardiology, 2021, 331, 208-209.	1.7	2
62	First Steps of Population Genomic Medicine in the Arrhythmia World: Pros and Cons. Circulation, 2022, 145, 892-895.	1.6	2
63	175â€…Aortopathy-causing mutations increase aortic stiffness in healthy individuals. Heart, 2015, 101, A99.1-A99.	2.9	1
64	A comparative study of mutation screening of sarcomeric genes (MYBPC3 , MYH7 , TNNT2) using single gene approach versus targeted gene panel next generation sequencing in a cohort of HCM patients in Egypt. Egyptian Journal of Medical Human Genetics, 2017, 18, 381-387.	1.0	1
65	121â€…Re-evaluating the genetic contribution of monogenic dilated cardiomyopathy. , 2019, , .		1
66	Research in understudied populations offers local and global insights into the genetics of hypertrophic cardiomyopathy. Polish Archives of Internal Medicine, 2020, 130, 76-78.	0.4	1
67	76â€…Comprehensive Assessment of Rare Genetic Variation in Dilated Cardiomyopathy Genes in Patients and Controls: Abstract 76 Table 1. Heart, 2015, 101, A41.2-A42.	2.9	0
68	142â€…Effects of Truncating Variants in Titin on Cardiac Phenotype and Left Ventricular Remodelling in Dilated Cardiomyopathy. Heart, 2016, 102, A102-A103.	2.9	0
69	003â€…Precise phenotyping with CMR identifies moderate alcohol consumption as an important phenotypic modifier of titin cardiomyopathy. Heart, 2017, 103, A2.2-A3.	2.9	0
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	Improving risk prediction in hypertrophic cardiomyopathy: the key role of Dutch founder variants. Netherlands Heart Journal, 2021, 29, 299-300.	0.8	0