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	Evaluation of gene validity for CPVT and short QT syndrome in sudden arrhythmic death. European Heart Journal, 2022, 43, 1500-1510.	2.2	57
2	Minor hypertrophic cardiomyopathy genes, major insights into the genetics of cardiomyopathies. Nature Reviews Cardiology, 2022, 19, 151-167.	13.7	50
3	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
4	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
5	First Steps of Population Genomic Medicine in the Arrhythmia World: Pros and Cons. Circulation, 2022, 145, 892-895.	1.6	2
6	Disease-specific variant pathogenicity prediction significantly improves variant interpretation in inherited cardiac conditions. Genetics in Medicine, 2021, 23, 69-79.	2.4	39
7	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
8	Illuminating the path from genetics to clinical outcome in Brugada syndrome. European Heart Journal, 2021, 42, 1091-1093.	2.2	12
9	Desmin variants in cardiomyopathies â€“ the hard yards in defining pathogenicity. International Journal of Cardiology, 2021, 331, 208-209.	1.7	2
10	Improving risk prediction in hypertrophic cardiomyopathy: the key role of Dutch founder variants. Netherlands Heart Journal, 2021, 29, 299-300.	0.8	0
11	Evidence-Based Assessment of Genes in Dilated Cardiomyopathy. Circulation, 2021, 144, 7-19.	1.6	213
12	Rare variants in KDR, encoding VEGF Receptor 2, are associated with tetralogy of Fallot. Genetics in Medicine, 2021, 23, 1952-1960.	2.4	7
13	Variant Intronic Enhancer Controls SCN10A-short Expression and Heart Conduction. Circulation, 2021, 144, 229-242.	1.6	20
14	ALPK3: a full spectrum cardiomyopathy gene?. European Heart Journal, 2021, 42, 3074-3077.	2.2	4
15	New Variant With a Previously Unrecognized Mechanism of Pathogenicity in Hypertrophic Cardiomyopathy. Circulation, 2021, 144, 754-757.	1.6	4
16	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
17	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
18	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

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19	The Egyptian Collaborative Cardiac Genomics (ECCO-GEN) Project: defining a healthy volunteer cohort. Npj Genomic Medicine, 2020, 5, 46.	3.8	5
20	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
21	When genetic burden reaches threshold. European Heart Journal, 2020, 41, 3849-3855.	2.2	40
22	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
23	An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. Human Mutation, 2020, 41, 1577-1587.	2.5	10
24	Common and rare susceptibility genetic variants predisposing to Brugada syndrome in Thailand. Heart Rhythm, 2020, 17, 2145-2153.	0.7	23
25	Advantages and Perils of Clinical Whole-Exome and Whole-Genome Sequencing in Cardiomyopathy. Cardiovascular Drugs and Therapy, 2020, 34, 241-253.	2.6	21
26	Reevaluating the Genetic Contribution of Monogenic Dilated Cardiomyopathy. Circulation, 2020, 141, 387-398.	1.6	148
27	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
28	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
29	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
30	Evaluating the Clinical Validity of Hypertrophic Cardiomyopathy Genes. Circulation Genomic and Precision Medicine, 2019, 12, e002460.	3.6	267
31	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
32	Genetic Variants Associated With Cancer Therapyâ€“Induced Cardiomyopathy. Circulation, 2019, 140, 31-41.	1.6	195
33	121â€...Re-evaluating the genetic contribution of monogenic dilated cardiomyopathy. , 2019, , .		1
34	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
35	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
36	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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37	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
38	CardioClassifier: disease- and gene-specific computational decision support for clinical genome interpretation. Genetics in Medicine, 2018, 20, 1246-1254.	2.4	75
39	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
40	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
41	Dilated Cardiomyopathy DueÂtoÂBLC2-Associated AthanogeneÂ3Â(BAG3)ÂMutations. Journal of the American College of Cardiology, 2018, 72, 2471-2481.	2.8	93
42	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
43	Genetic Etiology for Alcohol-Induced Cardiac Toxicity. Journal of the American College of Cardiology, 2018, 71, 2293-2302.	2.8	182
44	Defining the genetic architecture of hypertrophic cardiomyopathy: re-evaluating the role of non-sarcomeric genes. European Heart Journal, 2017, 38, ehw603.	2.2	142
45	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
46	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
47	Using high-resolution variant frequencies to empower clinical genome interpretation. Genetics in Medicine, 2017, 19, 1151-1158.	2.4	355
48	Issues and Challenges in Diagnostic Sequencing for Inherited Cardiac Conditions. Clinical Chemistry, 2017, 63, 116-128.	3.2	7
49	Phenotype and Clinical Outcomes of TitinÂCardiomyopathy. Journal of the American College of Cardiology, 2017, 70, 2264-2274.	2.8	86
50	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
51	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
52	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
53	Titin-truncating variants affect heart function in disease cohorts and the general population. Nature Genetics, 2017, 49, 46-53.	21.4	255
54	Recovery of Cardiac Function in Cardiomyopathy Caused by Titin Truncation. JAMA Cardiology, 2016, 1, 234.	6.1	30

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55	Moderate Physical Activity in Healthy Adults Is Associated With Cardiac Remodeling. Circulation: Cardiovascular Imaging, 2016, 9, .	2.6	40
56	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
57	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
58	Titin truncating mutations: A rare cause of dilated cardiomyopathy in the young. Progress in Pediatric Cardiology, 2016, 40, 41-45.	0.4	23
59	Development of a Comprehensive Sequencing Assay for Inherited Cardiac Condition Genes. Journal of Cardiovascular Translational Research, 2016, 9, 3-11.	2.4	80
60	175â€...Aortopathy-causing mutations increase aortic stiffness in healthy individuals. Heart, 2015, 101, A99.1-A99.	2.9	1
61	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
62	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
63	Bayesian models for syndrome- and gene-specific probabilities of novel variant pathogenicity. Genome Medicine, 2015, 7, 5.	8.2	22
64	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
65	Detection of mutations in KLHL3 and CUL3 in families with FHHt (familial hyperkalaemic) Tj ETQq1 1 0.784314 rgBT /Ov	4.3	49
66	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
67	NECTAR: a database of codon-centric missense variant annotations. Nucleic Acids Research, 2014, 42, D1013-D1019.	14.5	3
68	Towards Clinical Molecular Diagnosis of Inherited Cardiac Conditions: A Comparison of Bench-Top Genome DNA Sequencers. PLoS ONE, 2013, 8, e67744.	2.5	51
69	Paralogous annotation of disease-causing variants in long QT syndrome genes. Human Mutation, 2012, 33, 1188-1191.	2.5	44
70	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
71	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