

# Roddy Walsh

## List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

67

papers

2,708

citations

26

h-index

51

g-index

81

ext. papers

3,960

ext. citations

8.9

avg, IF

4.67

L-index

#	Paper	IF	Citations
67	Targeted therapies in genetic dilated and hypertrophic cardiomyopathies: From molecular mechanisms to therapeutic targets.. <i>European Journal of Heart Failure</i> , <b>2021</b> ,	12.3	2
66	Evidence-Based Assessment of Genes in Dilated Cardiomyopathy. <i>Circulation</i> , <b>2021</b> , 144, 7-19	16.7	34
65	Rare variants in KDR, encoding VEGF Receptor 2, are associated with tetralogy of Fallot. <i>Genetics in Medicine</i> , <b>2021</b> , 23, 1952-1960	8.1	1
64	Disease-specific variant pathogenicity prediction significantly improves variant interpretation in inherited cardiac conditions. <i>Genetics in Medicine</i> , <b>2021</b> , 23, 69-79	8.1	4
63	Enhancing rare variant interpretation in inherited arrhythmias through quantitative analysis of consortium disease cohorts and population controls. <i>Genetics in Medicine</i> , <b>2021</b> , 23, 47-58	8.1	13
62	Illuminating the path from genetics to clinical outcome in Brugada syndrome. <i>European Heart Journal</i> , <b>2021</b> , 42, 1091-1093	9.5	6
61	Variant Intronic Enhancer Controls Expression and Heart Conduction. <i>Circulation</i> , <b>2021</b> , 144, 229-242	16.7	1
60	New Variant With a Previously Unrecognized Mechanism of Pathogenicity in Hypertrophic Cardiomyopathy. <i>Circulation</i> , <b>2021</b> , 144, 754-757	16.7	1
59	Phenotypic Expression and Outcomes in Individuals With Rare Genetic Variants of Hypertrophic Cardiomyopathy. <i>Journal of the American College of Cardiology</i> , <b>2021</b> , 78, 1097-1110	15.1	3
58	Evaluation of gene validity for CPVT and short QT syndrome in sudden arrhythmic death. <i>European Heart Journal</i> , <b>2021</b> ,	9.5	5
57	Minor hypertrophic cardiomyopathy genes, major insights into the genetics of cardiomyopathies. <i>Nature Reviews Cardiology</i> , <b>2021</b> ,	14.8	5
56	Shared genetic pathways contribute to risk of hypertrophic and dilated cardiomyopathies with opposite directions of effect. <i>Nature Genetics</i> , <b>2021</b> , 53, 128-134	36.3	35
55	Systematic large-scale assessment of the genetic architecture of left ventricular noncompaction reveals diverse etiologies. <i>Genetics in Medicine</i> , <b>2021</b> , 23, 856-864	8.1	12
54	When genetic burden reaches threshold. <i>European Heart Journal</i> , <b>2020</b> , 41, 3849-3855	9.5	12
53	Transethnic Genome-Wide Association Study Provides Insights in the Genetic Architecture and Heritability of Long QT Syndrome. <i>Circulation</i> , <b>2020</b> , 142, 324-338	16.7	27
52	An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. <i>Human Mutation</i> , <b>2020</b> , 41, 1577-1587	4.7	4
51	Common and rare susceptibility genetic variants predisposing to Brugada syndrome in Thailand. <i>Heart Rhythm</i> , <b>2020</b> , 17, 2145-2153	6.7	8

50	Advantages and Perils of Clinical Whole-Exome and Whole-Genome Sequencing in Cardiomyopathy. <i>Cardiovascular Drugs and Therapy</i> , <b>2020</b> , 34, 241-253	3.9	13
49	Reevaluating the Genetic Contribution of Monogenic Dilated Cardiomyopathy. <i>Circulation</i> , <b>2020</b> , 141, 387-398	16.7	71
48	Contemporary Insights Into the Genetics of Hypertrophic Cardiomyopathy: Toward a New Era in Clinical Testing?. <i>Journal of the American Heart Association</i> , <b>2020</b> , 9, e015473	6	15
47	The Egyptian Collaborative Cardiac Genomics (ECCO-GEN) Project: defining a healthy volunteer cohort. <i>Npj Genomic Medicine</i> , <b>2020</b> , 5, 46	6.2	2
46	Genetic Studies of Hypertrophic Cardiomyopathy in Singaporeans Identify Variants in and That Are Common in Chinese Patients. <i>Circulation Genomic and Precision Medicine</i> , <b>2020</b> , 13, 424-434	5.2	4
45	Evaluating the Clinical Validity of Hypertrophic Cardiomyopathy Genes. <i>Circulation Genomic and Precision Medicine</i> , <b>2019</b> , 12, e002460	5.2	132
44	Quantitative approaches to variant classification increase the yield and precision of genetic testing in Mendelian diseases: the case of hypertrophic cardiomyopathy. <i>Genome Medicine</i> , <b>2019</b> , 11, 5	14.4	54
43	Genetic Variants Associated With Cancer Therapy-Induced Cardiomyopathy. <i>Circulation</i> , <b>2019</b> , 140, 31-41	16.7	110
42	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. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 284-292	8.1	32
41	Using High-Resolution Variant Frequencies Empowers Clinical Genome Interpretation and Enables Investigation of Genetic Architecture. <i>American Journal of Human Genetics</i> , <b>2019</b> , 104, 187-190	11	8
40	Analysis of 51 proposed hypertrophic cardiomyopathy genes from genome sequencing data in sarcomere negative cases has negligible diagnostic yield. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 1576-1584	8.1	25
39	CardioClassifier: disease- and gene-specific computational decision support for clinical genome interpretation. <i>Genetics in Medicine</i> , <b>2018</b> , 20, 1246-1254	8.1	45
38	Adaptation and validation of the ACMG/AMP variant classification framework for MYH7-associated inherited cardiomyopathies: recommendations by ClinGen's Inherited Cardiomyopathy Expert Panel. <i>Genetics in Medicine</i> , <b>2018</b> , 20, 351-359	8.1	173
37	Dilated Cardiomyopathy Due to BCL2-Associated Athanogene 3 (BAG3) Mutations. <i>Journal of the American College of Cardiology</i> , <b>2018</b> , 72, 2471-2481	15.1	53
36	Complex roads from genotype to phenotype in dilated cardiomyopathy: scientific update from the Working Group of Myocardial Function of the European Society of Cardiology. <i>Cardiovascular Research</i> , <b>2018</b> , 114, 1287-1303	9.9	57
35	Genetic Etiology for Alcohol-Induced Cardiac Toxicity. <i>Journal of the American College of Cardiology</i> , <b>2018</b> , 71, 2293-2302	15.1	112
34	Defining the genetic architecture of hypertrophic cardiomyopathy: re-evaluating the role of non-sarcomeric genes. <i>European Heart Journal</i> , <b>2017</b> , 38, 3461-3468	9.5	101
33	Histiocytoid cardiomyopathy and microphthalmia with linear skin defects syndrome: phenotypes linked by truncating variants in. <i>Journal of Physical Education and Sports Management</i> , <b>2017</b> , 3, a001271	2.8	15

32	Truncating Variants in Titin Independently Predict Early Arrhythmias in Patients With Dilated Cardiomyopathy. <i>Journal of the American College of Cardiology</i> , <b>2017</b> , 69, 2466-2468	15.1	40
31	Using high-resolution variant frequencies to empower clinical genome interpretation. <i>Genetics in Medicine</i> , <b>2017</b> , 19, 1151-1158	8.1	208
30	Issues and Challenges in Diagnostic Sequencing for Inherited Cardiac Conditions. <i>Clinical Chemistry</i> , <b>2017</b> , 63, 116-128	5.5	6
29	Phenotype and Clinical Outcomes of Titin Cardiomyopathy. <i>Journal of the American College of Cardiology</i> , <b>2017</b> , 70, 2264-2274	15.1	57
28	003 Precise phenotyping with CMR identifies moderate alcohol consumption as an important phenotypic modifier of titin cardiomyopathy. <i>Heart</i> , <b>2017</b> , 103, A2.2-A3	5.1	
27	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. <i>Egyptian Journal of Medical Human Genetics</i> , <b>2017</b> , 18, 381-387	2	1
26	Reassessment of Mendelian gene pathogenicity using 7,855 cardiomyopathy cases and 60,706 reference samples. <i>Genetics in Medicine</i> , <b>2017</b> , 19, 192-203	8.1	386
25	Titin-truncating variants affect heart function in disease cohorts and the general population. <i>Nature Genetics</i> , <b>2017</b> , 49, 46-53	36.3	179
24	Moderate Physical Activity in Healthy Adults Is Associated With Cardiac Remodeling. <i>Circulation: Cardiovascular Imaging</i> , <b>2016</b> , 9,	3.9	27
23	142 Effects of Truncating Variants in Titin on Cardiac Phenotype and Left Ventricular Remodelling in Dilated Cardiomyopathy. <i>Heart</i> , <b>2016</b> , 102, A102-A103	5.1	
22	143 Clinical and Genetic Characteristics of Familial Dilated Cardiomyopathy in a Large UK Prospective Cohort. <i>Heart</i> , <b>2016</b> , 102, A103-A104	5.1	2
21	Titin truncating mutations: A rare cause of dilated cardiomyopathy in the young. <i>Progress in Pediatric Cardiology</i> , <b>2016</b> , 40, 41-45	0.4	17
20	Development of a Comprehensive Sequencing Assay for Inherited Cardiac Condition Genes. <i>Journal of Cardiovascular Translational Research</i> , <b>2016</b> , 9, 3-11	3.3	63
19	Recovery of Cardiac Function in Cardiomyopathy Caused by Titin Truncation. <i>JAMA Cardiology</i> , <b>2016</b> , 1, 234-5	16.2	21
18	Bayesian models for syndrome- and gene-specific probabilities of novel variant pathogenicity. <i>Genome Medicine</i> , <b>2015</b> , 7, 5	14.4	19
17	Integrated allelic, transcriptional, and phenomic dissection of the cardiac effects of titin truncations in health and disease. <i>Science Translational Medicine</i> , <b>2015</b> , 7, 270ra6	17.5	267
16	175 Aortopathy-causing mutations increase aortic stiffness in healthy individuals. <i>Heart</i> , <b>2015</b> , 101, A99.1-A99 1	5.1	
15	Genetic modifiers to the PLN L39X mutation in a patient with DCM and sustained ventricular tachycardia?. <i>Global Cardiology Science &amp; Practice</i> , <b>2015</b> , 2015, 29	0.7	3

14	76 Comprehensive Assessment of Rare Genetic Variation in Dilated Cardiomyopathy Genes in Patients and Controls. <i>Heart</i> , <b>2015</b> , 101, A41.2-A42	5.1	
13	Detection of mutations in KLHL3 and CUL3 in families with FHt (familial hyperkalaemic hypertension or Gordon's syndrome). <i>Clinical Science</i> , <b>2014</b> , 126, 721-6	6.5	38
12	Paralogue annotation identifies novel pathogenic variants in patients with Brugada syndrome and catecholaminergic polymorphic ventricular tachycardia. <i>Journal of Medical Genetics</i> , <b>2014</b> , 51, 35-44	5.8	31
11	NECTAR: a database of codon-centric missense variant annotations. <i>Nucleic Acids Research</i> , <b>2014</b> , 42, D1013-9	20.1	2
10	Towards clinical molecular diagnosis of inherited cardiac conditions: a comparison of bench-top genome DNA sequencers. <i>PLoS ONE</i> , <b>2013</b> , 8, e67744	3.7	48
9	Paralogous annotation of disease-causing variants in long QT syndrome genes. <i>Human Mutation</i> , <b>2012</b> , 33, 1188-1191	4.7	26
8	Anti-inflammatory modulation of chronic airway inflammation in the murine house dust mite model. <i>Pulmonary Pharmacology and Therapeutics</i> , <b>2008</b> , 21, 637-47	3.5	35
7	Time course toxicogenomic profiles in CD-1 mice after nontoxic and nonlethal hepatotoxic paracetamol administration. <i>Chemical Research in Toxicology</i> , <b>2004</b> , 17, 1551-61	4	17
6	Quantitative approaches to variant classification increase the yield and precision of genetic testing in Mendelian diseases: The case of hypertrophic cardiomyopathy		1
5	Genomics of Egyptian Healthy Volunteers: The EHVol Study		1
4	Using high-resolution variant frequencies to empower clinical genome interpretation		4
3	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
2	Analysis of HCM in an understudied population reveals a new mechanism of pathogenicity		2
1	Outcomes and phenotypic expression of rare variants in hypertrophic cardiomyopathy genes amongst UK Biobank participants		1