Roddy Walsh

List of Publications by Citations

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	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
66	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
65	Using high-resolution variant frequencies to empower clinical genome interpretation. *Genetics in Medicine*, **2017**, 19, 1151-1158	8.1	208
64	Titin-truncating variants affect heart function in disease cohorts and the general population. *Nature Genetics*, **2017**, 49, 46-53	36.3	179
63	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
62	Evaluating the Clinical Validity of Hypertrophic Cardiomyopathy Genes. *Circulation Genomic and Precision Medicine*, **2019**, 12, e002460	5.2	132
61	Genetic Etiology for Alcohol-Induced Cardiac Toxicity. *Journal of the American College of Cardiology*, **2018**, 71, 2293-2302	15.1	112
60	Genetic Variants Associated With Cancer Therapy-Induced Cardiomyopathy. *Circulation*, **2019**, 140, 31-41	16.7	110
59	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
58	Reevaluating the Genetic Contribution of Monogenic Dilated Cardiomyopathy. *Circulation*, **2020**, 141, 387-398	16.7	71
57	Development of a Comprehensive Sequencing Assay for Inherited Cardiac Condition Genes. *Journal of Cardiovascular Translational Research*, **2016**, 9, 3-11	3.3	63
56	Phenotype and Clinical Outcomes of Titin Cardiomyopathy. *Journal of the American College of Cardiology*, **2017**, 70, 2264-2274	15.1	57
55	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	9.9	57
54	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
53	Dilated Cardiomyopathy Due to BLC2-Associated Athanogene 3 (BAG3) Mutations. *Journal of the American College of Cardiology*, **2018**, 72, 2471-2481	15.1	53
52	Towards clinical molecular diagnosis of inherited cardiac conditions: a comparison of bench-top genome DNA sequencers. *PLoS ONE*, **2013**, 8, e67744	3.7	48
51	CardioClassifier: disease- and gene-specific computational decision support for clinical genome interpretation. *Genetics in Medicine*, **2018**, 20, 1246-1254	8.1	45

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	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
48	Anti-inflammatory modulation of chronic airway inflammation in the murine house dust mite model. *Pulmonary Pharmacology and Therapeutics*, **2008**, 21, 637-47	3.5	35
47	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
46	Evidence-Based Assessment of Genes in Dilated Cardiomyopathy. *Circulation*, **2021**, 144, 7-19	16.7	34
45	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	8.1	32
44	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
43	Transethnic Genome-Wide Association Study Provides Insights in the Genetic Architecture and Heritability of Long QT Syndrome. *Circulation*, **2020**, 142, 324-338	16.7	27
42	Moderate Physical Activity in Healthy Adults Is Associated With Cardiac Remodeling. *Circulation: Cardiovascular Imaging*, **2016**, 9,	3.9	27
41	Paralogous annotation of disease-causing variants in long QT syndrome genes. *Human Mutation*, **2012**, 33, 1188-1191	4.7	26
40	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
39	Recovery of Cardiac Function in Cardiomyopathy Caused by Titin Truncation. *JAMA Cardiology*, **2016**, 1, 234-5	16.2	21
38	Bayesian models for syndrome- and gene-specific probabilities of novel variant pathogenicity. *Genome Medicine*, **2015**, 7, 5	14.4	19
37	Titin truncating mutations: A rare cause of dilated cardiomyopathy in the young. *Progress in Pediatric Cardiology*, **2016**, 40, 41-45	0.4	17
36	Time course toxicogenomic profiles in CD-1 mice after nontoxic and nonlethal hepatotoxic paracetamol administration. *Chemical Research in Toxicology*, **2004**, 17, 1551-61	4	17
35	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
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	6	15
33	Advantages and Perils of Clinical Whole-Exome and Whole-Genome Sequencing in Cardiomyopathy. *Cardiovascular Drugs and Therapy*, **2020**, 34, 241-253	3.9	13

32	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
31	When genetic burden reaches threshold. *European Heart Journal*, **2020**, 41, 3849-3855	9.5	12
30	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
29	Common and rare susceptibility genetic variants predisposing to Brugada syndrome in Thailand. *Heart Rhythm*, **2020**, 17, 2145-2153	6.7	8
28	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
27	Issues and Challenges in Diagnostic Sequencing for Inherited Cardiac Conditions. *Clinical Chemistry*, **2017**, 63, 116-128	5.5	6
26	Illuminating the path from genetics to clinical outcome in Brugada syndrome. *European Heart Journal*, **2021**, 42, 1091-1093	9.5	6
25	Evaluation of gene validity for CPVT and short QT syndrome in sudden arrhythmic death. *European Heart Journal*, **2021**,	9.5	5
24	Minor hypertrophic cardiomyopathy genes, major insights into the genetics of cardiomyopathies. *Nature Reviews Cardiology*, **2021**,	14.8	5
23	An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. *Human Mutation*, **2020**, 41, 1577-1587	4.7	4
22	Using high-resolution variant frequencies to empower clinical genome interpretation		4
21	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
20	Disease-specific variant pathogenicity prediction significantly improves variant interpretation in inherited cardiac conditions. *Genetics in Medicine*, **2021**, 23, 69-79	8.1	4
19	Genetic modifiers to the PLN L39X mutation in a patient with DCM and sustained ventricular tachycardia?. *Global Cardiology Science & Practice*, **2015**, 2015, 29	0.7	3
18	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
17	143 Clinical and Genetic Characteristics of Familial Dilated Cardiomyopathy in a Large UK Prospective Cohort. *Heart*, **2016**, 102, A103-A104	5.1	2
16	NECTAR: a database of codon-centric missense variant annotations. *Nucleic Acids Research*, **2014**, 42, D1013-9	20.1	2
15	Targeted therapies in genetic dilated and hypertrophic cardiomyopathies: From molecular mechanisms to therapeutic targets.. *European Journal of Heart Failure*, **2021**,	12.3	2

List of Publications

14	Analysis of HCM in an understudied population reveals a new mechanism of pathogenicity		2
13	The Egyptian Collaborative Cardiac Genomics (ECCO-GEN) Project: defining a healthy volunteer cohort. *Npj Genomic Medicine*, **2020**, 5, 46	6.2	2
12	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	2	1
11	175 Aortopathy-causing mutations increase aortic stiffness in healthy individuals. *Heart*, **2015**, 101, A99.1-A99	5.1	1
10	Quantitative approaches to variant classification increase the yield and precision of genetic testing in Mendelian diseases: The case of hypertrophic cardiomyopathy		1
9	Genomics of Egyptian Healthy Volunteers: The EHVol Study		1
8	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
7	Rare variants in KDR, encoding VEGF Receptor 2, are associated with tetralogy of Fallot. *Genetics in Medicine*, **2021**, 23, 1952-1960	8.1	1
6	Variant Intronic Enhancer Controls Expression and Heart Conduction. *Circulation*, **2021**, 144, 229-242	16.7	1
5	New Variant With a Previously Unrecognized Mechanism of Pathogenicity in Hypertrophic Cardiomyopathy. *Circulation*, **2021**, 144, 754-757	16.7	1
4	Outcomes and phenotypic expression of rare variants in hypertrophic cardiomyopathy genes amongst UK Biobank participants		1
3	142 Effects of Truncating Variants in Titin on Cardiac Phenotype and Left Ventricular Remodelling in Dilated Cardiomyopathy. *Heart*, **2016**, 102, A102-A103	5.1	
2	003 Precise phenotyping with CMR identifies moderate alcohol consumption as an important phenotypic modifier of titin cardiomyopathy. *Heart*, **2017**, 103, A2.2-A3	5.1	
1	76 Comprehensive Assessment of Rare Genetic Variation in Dilated Cardiomyopathy Genes in Patients and Controls. *Heart*, **2015**, 101, A41.2-A42	5.1	